Title

Immaculate Re-Conception: Redefining Health and Reproductive Risk Using Prenatal Genetic Testing

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Publication Date

2024

Peer reviewed|Thesis/dissertation

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By

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A dissertation submitted in partial satisfaction of the

requirements for the degree of

Doctor of Philosophy

in

Sociology

in the

Graduate Division

of the

University of California, Berkeley

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Professor Raka Ray, Chair
Professor Neil Fligstein
Professor Osagie Obasogie
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Spring 2024

ABSTRACT

Immaculate Re-Conception: Redefining Health and Reproductive Risk Using Prenatal Genetic Testing

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This dissertation explores the routinization of prenatal genetic testing in the United States' precision medicine landscape. It investigates how these technologies shape perspectives on reproduction, health, and disability, and probes the ethical implications of their widespread use. I ask, how does the "appropriate" use of prenatal genetic technologies come to frame certain existences as meaningful and worthwhile while others are seen as unwanted and less valuable? I also explore the role of experts in both implementing and interpreting these technologies during medical research as well as patientcare. The study reveals how the routine use of prenatal genetic technologies positions disability as an undesirable social harm, narrowing our tolerance of difference and amplifying the imperative to use these tools to reproduce 'judiciously.'

The first empirical chapter delves into Wrongful Birth and Life legal disputes in prenatal testing. It reveals how courts have ushered the technological imperative around prenatal genetic tools and enshrined disability as an unwanted private burden. In the second, I unpack the social making of genetic diagnostic categories. Focusing on sex chromosome aneuploidies, this chapter offers an in-depth examination of gendered pathology and the medicalization of genetic variations as 'abnormal' existences. The final empirical chapter centers on experts and expertise. It illuminates how reproductive physicians and genetic counselors organize expertise and responsibilities around prenatal testing, emphasizing possible transformations in which groups are influencing the forefront of genomics and equitable patientcare.

I employ mixed qualitative methods. I conducted in-depth interviews (20 with reproductive physicians, 20 with genetic counselors, and 20 with patients). Further, I completed ethnographic observations at professional genomics conferences, a genetic counseling master's program, and consultations between patients and genetic counselors. I also relied on content analysis, using a Python-assisted web scraper to gather text-based exchanges about prenatal genetic testing on Reddit. Finally, I conducted archival research on Wrongful Birth and Life cases from 1963-2021.

There are significant consequences surrounding genetic technologies, systematic de-selection of disability, reproductive pressures, and the enduring history of eugenics. As prenatal genetic innovations become more precise, capable, and accessible, it is essential to implement them toward a more inclusive and just society. To be sure, prenatal genetic testing is not inherently harmful; rather, harms result from how we systematically employ these tools to treat disability as objectively unwanted. In that vein, this dissertation is an endeavor to pave the way for a more equitable and compassionate future in prenatal testing, where inclusive social infrastructures parallel technological innovation.

ACKNOWLEDGEMENTS

This dissertation would not exist without the insightfulness and generosity of all those who participated. I am grateful for the clinics and genetic counseling programs that welcomed me into their space. Each genetic counselor, physician, and parent interviewed was so gracious with their time and perspectives. Often, these interviews were emotionally challenging and brought up complex issues – I appreciate you all for exploring this terrain with me.

To Raka Ray, my advisor, chair, and so much more, *ki bolbo*? From my first year, you made me feel not only like I belong but insisted that I be ambitious. The community of feminist scholars you nurtured was an unparalleled source of support. Thank you for seeing me as a whole person, cultivating in me a scholar, and encouraging me to lead from my standpoint. In my toughest times, while I wish it had not been, you showed me the path to resilience. I am forever fortunate to have been your student.

To my committee members Neil Fligstein, Osagie Obasogie, and Daniel Navon, I have learned volumes from each of you. You have taught me to sharpen my thinking and be rigorous with my work. Through countless meetings and marked-up documents, you have been generous with your time and knowledge. I am most grateful for the compassion and grace you showed me during my final mile, which was made so much more trying than I could have imagined.

I have had the privilege of learning from the most outstanding faculty mentors and graduate students. Ann Keller, Eliza Brown, Danya Lagos, Robert Braun, and Armando Lara-Millán, thank you for guiding me, giving me crucial opportunities, and supporting me beyond my work. Catherine Norton, I am appreciative for the time, patience, and kindness you commit to helping us navigate each aspect of being a graduate student. I have also benefited greatly from the camaraderie and support among our graduate students. To everyone who dedicated their time to the SWAG and GWNFSM writing groups, thank you! These chapters would not exist without your steady solidarity. I am also grateful to everyone in the 2017 cohort, Gender and Sexuality Working Group, Berkeley Connect Fellows Working Group, and Raka's Dissertation Group. I have been so enriched by your perspectives and friendship.

Zaina Mahmoud, I am especially thankful to you for persevering through our Wrongful Birth and Life project. From strangers on a mailing list, to overseas co-authors, you have become a friend above all. Natalie Rivas, Karen Tirado, and Elizabeth Brown, I am so lucky to have had each of you as research assistants and mentees. This dissertation would not be where it is without your passion, research efforts, and thoughtfulness.

This work has been enabled by several research institutes. The Institute for Citizens and Scholars, UC Berkeley's Institute for South Asia Studies, and Center for Race and Gender provided generous fellowships and grants. I have also benefited from resources and intellectual community within UC Berkeley's Institute for the Study of Societal Issues, Institute for Research on Labor and Employment, and the Reproscholars Network.

Finally, where would I be without the relentless encouragement from my loving and ever-optimistic family? Mama and Dad, you have given me the greatest gifts: endless curiosity, confidence, and a drive to care. Kiki, you have been proof-reading and transcribing for me since you could type – I owe you. Dadu and Didun, I am indebted to you for instilling in me your passion for social equity, storytelling, and a love for all things scholarly. Dadubhai, Didibhai, I know how proud this would have made you.

Rohan, from staying awake during my writing nights to listening as I ramble, you have seen me through each step. You nurtured me when I was exhausted and pushed me when the time was right. In the face of a seemingly insurmountable obstacle, you were beside me until I could take the reins. You and Bean have given these past years the balance they needed; it is our constant adventures and mundane joys that made this process that much more possible and fulfilling.

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CHAPTER ONE

Introduction

Embarking on the Precision Medicine Era

"We're here to harness what is most special about America, and that is our spirit of innovation; our ability to dream and take risks, and tinker and try new things. And as a result of that, it will not only improve our economy, but improve the lives of men and women and children for generations to come. And together, what's so exciting is […] that we have the possibility of leading an entirely new era of medicine that makes sure new jobs and new industries and new lifesaving treatments for diseases are created right here in the United States (Obama 2015)."

These were President Barack Obama's words in his January 2015 State of the Union Address, where he formally ushered in the Precision Medicine Initiative (PMI) – a new era in medicine, bringing with it new promises. The initiative committed to building out the precision or personalized medicine industry in the United States, developing more medical technologies that take an individualized approach to managing disease based on a person's genetics, environment, and lifestyle. With precision medicine tools, scientists and doctors sought to revolutionize how we approach health and risk management, using personalized data about genetic biomarkers, epidemiological factors, and individuals' life circumstances to better understand a person's health needs, risks, and effective treatments (Phillips 2020). Precision medicine would allow us to address health on a personalized level, diagnosing conditions and diseases more quickly and implementing treatments and preventative measures specifically suited for each patient.

These innovations were framed not only as a health benefit but also as an economic driver in terms of jobs created and industries fostered. President Obama went on to discuss this "promise of precision medicine," assuring the public that these technologies would deliver "the right treatments, at the right time, every time to the right person." He underscored the power and, more importantly, the economic stakes of genetic technologies as "one of the greatest opportunities for new medical breakthroughs that we have *ever* seen," highlighting that for every dollar spent on the Human Genome Project – the endeavor to map the entire human genome – it had returned $140 to the United States economy. Before closing, President Obama called forward an era of technologized and individualized medicine, harkening back to genomics developments that paved the way, "The time is right to unleash a new wave of advances in this area, in precision medicine, just like we did with genetics 25 years ago." (Obama 2015; White House 2015b)

Since it was founded, the Precision Medicine Initiative has received over $6.5 billion in funding from the United States government and private sources. A year after it was set into motion, PMI received $215 million in the President's 2016 Budget. In years following, PMI has been allocated over $5.5 billion across the National Institutes of Health's 2017-2020 and 2021-2024 budgets, and $700 million from the Food and Drug Administration. It has also received robust funding from private foundations, like the Bill and Melinda Gates Foundation's $100 million contribution, and from industry partners, such as the pharmaceutical company Pfizer that committed $50 million (Advisory Committee 2018; Collins and Varmus 2015; National Institutes of Health 2022; Sarata and Johnson 2016; White House 2015a).

Among these shiny new tools of promise to receive massive investments are new genetic technologies – arguably, the core of the precision medicine era. Today, genetic technologies are intertwined with how we understand ourselves and approach medical treatments and health management. They have carved out a direct-to-consumer testing market and have been instrumental in the diagnosis, treatment, and prevention of diseases in fields such as cancer, pediatrics, and reproduction, where they are routinely implemented (Lippman 1992; Mukherjee et al. 2022; Navon 2019; Nelson 2018; Roberts 2012; Rose 2001). Reproduction has become a key setting for the innovation and implementation of these tools. Since the 1950s, new reproductive technologies have paralleled breakthrough findings in genomics; as geneticists began uncovering gene biomarkers underlying various health conditions during this time, scientists applied these findings to create amniocentesis, the first prenatal genetic diagnostic technology (Minna Stern 2012; National Institutes of Health n.d.). Because reproductive genetic technologies enable selection against particular conditions and diseases, they have been heralded as an essential health and pregnancy management solution within medical institutions and often incorporated into public health measures for population health.

These tools, however, are not a silver bullet to our growing health needs, and have significant implications for how we approach disability, reproduction, and broader social inclusivity. As such, they need to be studied more carefully and critically before they are widely implemented. In this dissertation, I focus on genetic technologies as they are used during pregnancy and reproduction – namely, prenatal genetic technologies that are used to test for fetal genetic conditions. I explore how these prenatal genetic technologies have been used over time and how they have shaped our understandings of disability, health and risk management, reproductive choices, and even social equity. Throughout, I emphasize the ongoing need to re-envision what it means to use these tools toward securing 'health' and 'normalcy,' situating these genetic developments within their undeniably harmful histories and social inequities.

In this introductory chapter, I begin by discussing the history of eugenics that grounds contemporary precision medicine, especially in how it bears on reproductive genetics. Next, I provide a historical overview of reproductive genetic technology development, bringing us to the contemporary landscape of widespread Non-Invasive Prenatal Testing (NIPT) use. Then, I discuss the key experts who are involved in developing and implementing these tools in today's prenatal testing space, focusing on reproductive physicians, geneticists, and genetic counselors. Following this, I delve into the emerging issues in prenatal genetic testing, which inform the central questions and methods employed in this dissertation. I focus on three emerging issues: the pressures to pursue prenatal genetic testing (i.e., technological imperatives), the backdrop of eugenics that persists in medicalized testing practices, and the implications that routinized prenatal genetic testing has on shaping understandings of disability and meaningful existence. Finally, I present an overview of the three empirical chapters comprising this dissertation.

The Ghosts of Eugenics

While genetic technologies and the science enabling them may seem like a flashy innovation of the contemporary personalized medicine era, genetics has been a part of public health and medicine since the early 1900s. Moreover, genetics has always been closely tied to family-building and reproduction. Early geneticists (many of whom were considered eugenicists) regularly published about population control and family-planning, seeking to control the fertility of non-white families

and disabled individuals. In its earliest forms, genetics was often employed in 'heredity clinics,' where social workers and health professionals evaluated Black and/or disabled orphans to understand whether they could socially pass as adoptive members of white families (Minna Stern 2012). In its deadliest forms, genetics has been used to further the eugenics agenda grounded in white and ableist supremacy (Black 2003; Obasogie and Darnovsky 2018). Despite its troubled history, genetics is an essential part of contemporary medicine with a firm base in reproduction-related fields, having enabled critical live-saving treatments and provided individuals with greater control over their health outcomes. To harness their insights and potential thoughtfully – with an eye toward social equity – it is critical to center the historical context of genetic technologies, especially their application to reproduction.

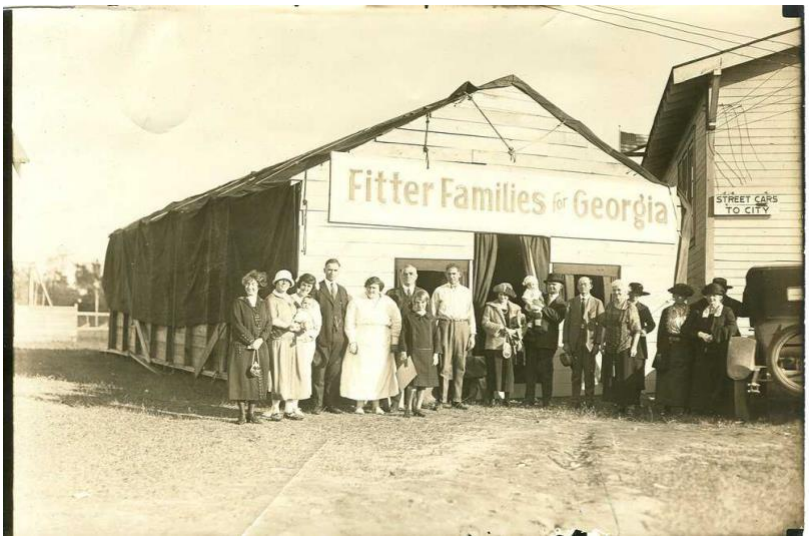
History often associates eugenics with Nazism. However, eugenics finds its roots much earlier across the United States and Europe. Biostatisticians in the early 1900s – including Francis Galton, cousin of the famous evolutionary scientist Charles Darwin – were deeply interested in making connections between heredity and disease in ways that created social hierarchies. They saw those with diseases and disabilities as genetically inferior, and often combined these ideas with racism to frame certain existences as superior to others. Using early foundations of statistics from the 1850s, biostatisticians who espoused eugenic values turned to the bell curve – the idea of a norm or average and outliers or deviations – to support their biases: "The norm pins down that majority of the population that falls under the arch of the standard bell-shaped curve. This curve, […] 'the bell curve,' became in its own way a symbol of tyranny of the norm. […] So, with the concept of the norm comes the concept of deviations or extremes. When we think of bodies, […] people with disabilities will be thought of as deviants" (Davis 1997:13). When statistics is applied to reign over the body, those within the 'normal' distribution of human features represented acceptable existences while those that strayed were abnormal deviations.[1] (Davis 1997; Obasogie and Darnovsky 2018)

Moving forward, throughout the 1930s and 1940s, the use of genetics toward eugenics principles of white supremacy, class-based privilege, and ableism became far clearer through institutionalization – solidifying the marriage between genetics and reproduction. Starting in the 1920s and 1930s, State Fairs across the United States conducted "Fitter Families" and "Better Babies" contests, where families and babies were "weighed, measured, and tested at the state fair by physicians and psychologists affiliated with the State Board of Health's Division of Infant and Child Hygiene" (Kline 2018; Minna Stern 2012; Obasogie and Darnovsky 2018). Throughout the 1930s and 1940s, hundreds of genetics clinics and related university departments sprung up around the United States, with geneticists of the time regularly lecturing and publishing about population control and

[1] When applied to the eugenics agenda, the bell curve, with its mean and outliers, catered to the idea of 'population betterment,' where societies ought to strive to reproduce and resemble normative ways of being. In public health, disability, and medicine, the bell curve was used to determine the 'normal' distribution of human features, representing desirable types of existence, while those who strayed from the average were seen as pathological, abnormal, and deviant (Davis 1997:11–13). Notions of normalcy and disability, thus, became understood as embodied and biologically essentialized, rather than products of social concepts and structures. Today, one might see semblances of these concepts in a pediatrician's office, to measure 'normal' child development, or with a primary physician when discussing bodily measures like height and weight. Though these metrics are not explicitly eugenics-oriented today, they do illustrate the ways in which statistics have made their way into defining normal and acceptable existence. (Obasogie and Darnovsky 2018)

family-planning "with an eye toward regulating fertility patterns" (Minna Stern 2012:57–58). One of the first of these genetics clinics was the Dight Institute in Minnesota, founded by Charles Dight – a famous eugenicist and strong proponent forced sterilization laws. These clinics were also home to the first genetic counseling practices, as the term 'genetic counseling' itself was coined in 1947 by Sheldon Reed, the Director of Dight Institute (Minna Stern 2012:17). Wickliffe Draper's Pioneer Fund largely underwrote these early genetics clinics. Draper was a "reclusive millionaire" who admired Hitler and Nazi policies. He aligned himself with scientists who believed Black people were biologically inferior, and in the early 1940s began investing in these newly founded medical genetics clinics (Minna Stern 2012:59–60). In exchange for his generous funding, these genetics clinics were to embody Draper's racist and ableist visions in their medical and scientific practice. They advocated for eugenics through anti-miscegenation practices and promoted sterilization as a "therapeutic weapon" (Minna Stern 2012:61). The agenda to weaponize genetics to control reproduction along discriminatory lines was steadily yet clearly taking hold in the United States.[2] (Black 2003; Obasogie and Darnovsky 2018)

A 'Fitter Family' contest at the Georgia State Fair in 1924. *Credit: American Philosophical Society, Eugenics Record Office Records.* (Kline 2018)

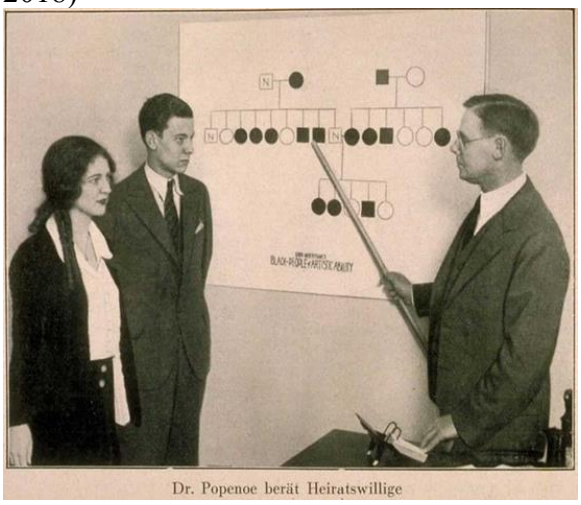


The main goal of early genetics centers in the United States was to conduct eugenics research and practice "genetic hygiene" techniques, where they educated and encouraged middle-class, white, able-bodied families to reproduce based on family pedigree charts. Some of these centers became known as 'heredity clinics' which worked with genetics clinics and research universities to produce pedigree carts tracing genetic markers of race, illness, and disability. During the 1940s, through the 1960s, these heredity clinics would adjudicate adoption cases for children of color. "Heredity clinics were inundated with requests from welfare and children's agencies to evaluate infants for skin color, racial characteristics, medical problems, and the likelihood of a genetic disorder" because adoption agencies "wanted to know about a newborn's eventual ability to convincingly

[2] It was not until the early 2000s that Wake Forest University, one of the first heredity clinics, "apologized for its eugenics program and for accepting Draper's money" (Minna Stern 2012). In October 2020 the University of California Berkeley also issued an apology for its Genealogical Eugenic Institute Fund, which was used to directly support the university's genetic counseling program founded in 1973 (Watanabe 2020).

appear white in American society" (Minna Stern 2012:44, 63). Akin to physiognomy – a now discredited pseudoscience – early geneticists and genetic counselors would judge infants based on racialized traits such as lip thickness, hair shape and texture, an eye fold for 'Mongolism,' colored smudges or pigments on the backs of finger joints, or a sacral spot at the base of the spine. They frequently employed University of California Berkeley geneticist Curt Stern's hypothesis about skin color genes, where normality was defined as closer to Caucasian presentation (Minna Stern 2012:64, 71). Accordingly, early medical genetics as employed in the United States was explicitly grounded in reproduction and eugenics, to support the State's population agenda of weeding out 'bad bloodlines' and 'feeble-mindedness' through regulating fertility and enforcing sterilizations. Women – typically those who were low-income, of color, or disabled – were forcibly sterilized, in part supporting the State's agenda to selectively reproduce those who resembled 'normalcy' and privilege along race, class, and health lines (Minna Stern 2012:20, 35–36). (Obasogie and Darnovsky 2018)

At a heredity clinic in 1930, eugenicist and doctor, Paul Popenoe shows a white couple a pedigree chart titled "Black People of Artistic Ability*." Credit: Cold Spring Harbor Laboratory.* (Kline 2018)



Dr. Popenoe berät Heiratswillige

After the Nazi-led horrors during World War Two, in the 1940s, the tone around genetics as used toward eugenics began to shift. Terminology around 'eugenics' gradually became taboo and faded, and with it went the explicit State-endorsed programs that privileged white supremacy and ableism. However, rather than being State-run, these practices around reproducing racial and class privilege, normative health, and able bodied-ness were steadily incorporated into social pressures on individuals to exercise their 'procreative autonomy' to reproduce 'responsibly' (which scholars often term 'liberal eugenics') (Fox 2012, 2018; Mills 2013). Key to this shift was the understanding of genetic markers that lead to various conditions occurring alongside the development of reproductive genetic technologies to test for these variations. Individuals could now be made responsible for using such tools when procreating and managing health risk; State-based programs could be cloaked in values of procreative autonomy to use reproductive genetic technologies.

In the 1950s and 1960s, scientists uncovered major findings in genomics research. They learned that humans are typically born with 46 chromosomes, that an extra 21st chromosome causes Down Syndrome. They learned how to genetically identify several conditions, including Turner

Syndrome, Klinefelter Syndrome, and Tay-Sachs. Around the world, geneticists were understanding the gene markers behind hundreds of "chromosomal anomalies" and developing technologies to visualize underlying genetic disease processes. In the United States, the National Institutes of Health played a major role in funding these advancements in genomic medicine and technologies, elevating a genetically essentialized and biologically determined understanding of health and ability while largely obscuring attention to social causes of entrenched health disparities (National Institutes of Health n.d.; Phillips 2020). These principles were reflected when scientists built reproductive genetic technologies of the time, including in vitro fertilization (IVF) (which led to the birth of Louise Brown in 1978); Robert G. Edwards, the IVF pioneer, was a staunch eugenicist and intended this tool – the staple technology of today's lucrative assisted reproduction industry – to better the "human genetic stock" (Obasogie 2013; Obasogie and Darnovsky 2018). Even though these critical findings and technological innovations lay the foundation for reproductive genomic medicine as an important therapeutic field, they bore 'eugenics ghosts' as scientists and doctors continued underscore that the purpose of genetics was to prevent disability and difference, foremost essentializing health to the body. (Minna Stern 2012; Navon 2019; Obasogie and Darnovsky 2018)

The remnants of eugenics lurk behind prenatal genetic technologies, as these tools were concurrently innovated alongside discoveries of genetic markers. When prenatal technologies such as amniocentesis and ultrasound took hold in the 1950s and 1970s, they were framed as tools to select against disability, as a morally appropriate and a cost-saving boon (Minna Stern 2012; Piepmeier 2013, 2015; Rapp 1994, 1998; R. Rapp 2001). These technologies were heralded for their potential to produce 'normal' able-bodied individuals and "save millions of dollars by reducing rates of institutionalization and producing more industrious citizens with higher overall earning power" (Minna Stern 2012:25). Feminist disabilities scholar Garland-Thomson has referred to this potential of reproductive technologies as "velvet eugenics," "for the soft, subtle way it encourages the eradication of disability" (Galis 2011; Garland-Thomson 2015). At this same time, however, the harmful historical agendas around genetics and reproduction were also becoming clearer for the public. For instance, the "slew of medical abuses linked to racism and population control policy" came to light in the 1970s, as women of color or poor white women who were forcibly sterilized made newspaper headlines and appeared in Congressional hearings and courtrooms to seek justice for the harms committed against them (Minna Stern 2012:72–73). This greater awareness around the misuses of genetics opens up a fruitful avenue for scholars, activists, and journalists to contextualize reproductive genetic technologies in their broader sociohistorical dynamics, potential for perpetuating racist and ableist ideas, and use toward essentializing an embodied understanding of health (Black 2003; Garland-Thomson 2015; Greely 2016; Minna Stern 2012; Obasogie 2013; Obasogie and Darnovsky 2018; Piepmeier 2015; Rapp 1994, 1998; R. Rapp 2001; Roberts 2012; Timmermans and Kaufman 2020; Watanabe 2020). The following chapters aim to situate prenatal genetic testing along these lines, arguing that the advantages these technologies bring can be best realized if implemented critically, with an understanding of their historical legacies.

Reproductive Genetic Technologies and the Non-Invasive Prenatal Testing 'Game Changer'

The medical field of reproduction has long been a budding ground for innovating and implementing genetic technologies. Embryos can be genetically tested using preimplantation genetic testing (PGT) for various conditions or typical development before implantation. Gamete

donors and hopeful parents often undergo carrier screening to test for conditions for which they may carry genes. And, using prenatal testing, pregnant patients can choose to genetically test their pregnancies to see if their fetus has a high probability for particular genetic conditions, confirm a genetic diagnosis, or sequence their fetus' genome or exome.[3] Prenatal genetic testing tools not only provide providers with a clearer understanding of how to care for a pregnancy given fetal health risks, but (in places where termination is permitted) they also allow expecting parents greater reproductive choice in which pregnancies they want to bring to term and how they manage their family's health. However, the rapidly routinized and systematic use of these prenatal genetic technologies has been criticized for its impact on stigmatizing disability via pressures to select against fetuses with chromosomal variations (e.g., Down Syndrome) – perhaps echoing a new, liberal eugenics (Fox 2012, 2018; Meredith et al. 2023; Mills 2013; Navon 2019). (Minear et al. 2015; National Institutes of Health n.d.; Piepmeier 2013, 2015; Rapp 1994, 1998; Zhang 2020)

One of the first prenatal genetic tests to emerge was amniocentesis, which was developed and implemented in clinics between the 1950s and 1970s; using a long needle inserted through the abdomen into the uterus to sample amniotic fluid, the technology enabled sequencing of fetal chromosomes to diagnose genetic conditions. In the 1980s, chorionic villus sampling (CVS), another invasive diagnostic test, and Maternal Serum Alpha Fetoprotein screening (MSAFP), which allowed for genetic screening via a maternal blood draw (testing fetal DNA in the maternal bloodstream), made their way into clinics (Minna Stern 2012). Throughout the 1970s and 1980s, these genetic technologies became widely used in clinical practice. In fact, by the end of the 1980s MSAFP and ultrasound was the standard of care in pregnancy management within the United States. By the end of the 1990s, it was also routine practice to screen pregnancies for Down Syndrome (trisomy 21) and other chromosomal variations within the first trimester, using tests for nuchal translucency (measures the fluid behind the fetus' neck) and ultrasounds. Over time, using insights from these genetic tests, doctors and scientists were also able to categorize pregnancies per their risk thresholds for developing congenital and genetic conditions, which led to more guidelines around testing for pregnant people. Where prenatal testing was previously reserved for pregnant people above the age of 35 or those with family histories of certain conditions, in 2007, the American College of Obstetricians and Gynecologists (ACOG) recommended that "prenatal screening using ultrasound, serum makers, and invasive prenatal testing be offered to all women, regardless of age" (Pergament 2013:63–66). This set the stage for almost effortlessly routinizing NIPT (or cell-free DNA testing) once it became available in 2011.

[3] There are two main types of prenatal genetic testing: screening and diagnostic. Screening (e.g., Non-invasive prenatal screening 'NIPT' and Maternal Serum Alpha Fetoprotein 'MSAFP') can identity whether a fetus is more or less likely to have certain congenital or genetic issues. Results are presented in terms of risk or probability that a fetus has particular conditions, including chromosomal conditions (e.g. trisomy 21 or 18), neural tube defects ("abnormalities of the brain or spinal cord"), sex chromosome aneuploidies, and the fetus' rhesus (Rh) blood type (which can severely complicate a pregnancy). Screening results, however, are not a diagnosis. They must be confirmed by a prenatal diagnostic test. Further, while screenings are typically non-invasive, many prenatal diagnostic tests are invasive. Diagnostic tests include chorionic villus sampling (CVS) and amniocentesis. They can confirm with 99% accuracy whether a fetus has trisomy 21 or 18, and with 90% accuracy whether a fetus has an open neural tube defect. Diagnostic tests can also identify other genetic disorders when targeted accordingly, often based on a patient's medical and obstetrics history. Given their ability to identify genetic conditions in-utero, prenatal genetic technologies have become a critical part of pregnancy management and family-building. (Biesecker 1998; California Legislature n.d.; Illumina 2023; Integrated Genetics 2020; Minear et al. 2015; Pergament 2013; Pös, Budiš, and Szemes 2019; Ravitsky et al. 2021)

Undoubtedly, NIPT has made prenatal genetic testing easier and less risky than ever. As the National Council on Disability underscores, "It would be hard to overstate the degree to which NIP(T) has altered how genetic testing and analysis is done" (National Council on Disability 2019:37). Released into the market around 2011, NIPT soon became a staple of pregnancy and healthcare in the United States. Not only do obstetrician gynecologists (ObGyns) today refer every pregnant patient to NIPT as standard practice, but ACOG describes itself as "engaged in proactive advocacy for NIPT coverage." At this point, it is estimated that only 1% of insurance providers *do not* cover NIPT, with the vast majority of major insurance providers, including Medicaid, covering the screening to some degree. Despite being widely accessible and routinely implemented, the test is not without its challenges. Fundamentally, it is a screening tool, so findings must be confirmed via diagnostic testing, and thorough genetic counseling is needed to explain the risk- or probability-based findings to patients (NIPT results are framed as the likelihood a fetus has particular genetic conditions tested for, which can be complicated for some patients and providers to grasp). There is also the possibility for false positives and false negatives, as well as inconclusive results, which may require repeat or follow-up testing. Still, experts in the field highlight that these challenges are outweighed by NIPT's supposed 'game changing' benefits when it comes to prenatal care. (American College of Obstetricians and Gynecologists 2021; Graham 2007; Pergament and Ilijic 2014)

There are several key components that set NIPT apart from its predecessors in prenatal genetic screening (and enable its rapid routinization). First, the test is low-risk and relatively straightforward to implement; it relies on a standard blood draw from the pregnant person and can be conducted as part of one's routine pregnancy blood work. Indeed, NIPT has a reputation of being 'just the blood draw' or 'the easy test' – a main selling point for genomics companies marketing this screening technology.[4] In the face of a 'long thin needle,' the option for a blood draw to receive a baseline of fetal genetic information without risking fetal loss is more appealing for many patients, psychologically and psychically (National Council on Disability 2019) (Genetic Counseling Ethnography 2021).

Second, NIPT screens for a range of common fetal genetic conditions and does so with relative accuracy, establishing it as a reliable testing tool. NIPT screens for Down Syndrome (trisomy 21), Edward Syndrome (trisomy 18), and Patau syndrome (trisomy 13), as well as sex chromosome aneuploidies such as Turner Syndrome (45,X), Klinefelter Syndrome (47,XYY), and Triple X Syndrome (47,XXX). Its sensitivity in identifying these common chromosomal conditions is quite high (certainly higher than other prenatal screenings), with 99.7%, 97.9%, and 99.0% for Down, Edward, and Patau syndromes, respectively, and 95.8% for Turner Syndrome (American College of Obstetricians and Gynecologists 2021; Ravitsky et al. 2021). Being able to rely on a non-invasive test to understand whether a fetus is affected by genetic conditions is informative for pregnancy management and care, including when it comes to pursuing appropriate treatments in-utero or during a child's early life, opting for further testing, or making decisions to terminate a pregnancy.

[4] Highlighting its non-invasiveness as a boon, providers often frame NIPT in relief to amniocentesis and CVS, which are invasive diagnostic tests, and thus pose a risk for miscarriage and other complications (though, providers cite that, today, chances of such issues are fewer than 1 in 1000) (Genetic Counseling Ethnography 2021).

Finally, NIPT can give pregnant patients and providers genetic information about a fetus' health (and sex) early in a pregnancy, as of 9 weeks, which is framed as an advantage for several reasons. For one, being able to identify sex chromosomes, NIPT has been vastly marketed as 'the gender test' to expecting parents who are eager to learn whether they are having a 'boy or girl' earlier in their pregnancy. This is a powerful selling point from genomics companies. Further, and more relevant to medical care, NIPT allows providers and expecting parents to understand the fetus' health state earlier than many other screenings. This facilitates timely clinical care or early pregnancy termination in cases where this is a desired and accessible intervention (Minear et al. 2015; National Council on Disability 2019). Given its use-value, as of 2020 ACOG recommends that reproductive providers offer NIPT to all pregnant people – regardless of age or baseline risk – illustrating how this technology marks a milestone in prenatal genetic testing, readily taken up into standard pregnancy care only a few years after its market release (American College of Obstetricians and Gynecologists 2021; Pergament and Ilijic 2014).

California, in particular, has a ripe landscape for routinizing prenatal genetic testing via NIPT, as the state has been running a publicly available Prenatal Screening Program since 1986. Seeking to make prenatal testing widely accessible state-wide, California offers pregnant people various types of prenatal screening, now including NIPT, for at most $232. The state's program also provides free follow-up services at approved Prenatal Diagnosis Centers, which can perform diagnostic tests, NIPT, ultrasounds, and provide genetic counseling. As of 2013, about 400,000 pregnant patients were participating in the state's screening program each year, representing around 75% of California's pregnant women. Of these pregnant patients, 77% of those who were at higher risk for their fetus having Down Syndrome accepted a referral to a diagnostic center, where they could access further testing and pregnancy interventions. As such, while these robust prenatal genetic testing options are voluntary for pregnant patients, they are encouraged and widely used as they are built into the state's public health priorities.[5] (California Department of Public Health 2023; California Legislature n.d.; Davis 1997; Flessel and Lorey 2011; Pergament and Ilijic 2014)

Unsurprisingly, key to NIPT's success in the United States is also the genomics companies that developed and marketed the technology.[6] While NIPT is not a direct-to-consumer genetic testing

[5] In addition to NIPT, which was recently incorporated into these offerings, the state provides pregnant people with quad marker screening (maternal blood sample at 15-20 weeks of pregnancy), serum integrated screening (combines a first and second trimester maternal blood test), and sequential integrated screening (combines first and second trimester maternal blood tests with nuchal translucency ultrasound test. Through these screens, pregnant patients can learn if their fetus is at a higher risk for open neural tube defects, abdominal wall defects, Down Syndrome (trisomy 21), Trisomy 13 and 18 (Patau and Edward Syndromes, respectively), and SLOS (Smith-Lemli-Opitz Syndrome). (California Department of Public Health 2023; California Legislature n.d.; Davis 1997; Flessel and Lorey 2011; Pergament and Ilijic 2014)

[6] Beyond commercial marketing strategies, these genomics companies have been at the heart of pushing forward NIPT routinization on a political and legislative front. In 2019, companies including Illumina, Myriad Women's Health, Labcorp, and Natera, formed the Coalition for Access to Prenatal Screening (CAPS), a lobbying organization that "seeks to improve access to state-of-the-art prenatal screening using (cell free) DNA-based NIP(T) that is easily accessible to all pregnant women who choose to pursue aneuploidy screening, regardless of their risk factors, income, age or geographic location" (Coalition for Access to Prenatal Screening 2023; National Council on Disability 2019). CAPS focuses on lobbying Congress for increasing Medicare and Medicaid funds to support NIPT, actively conducting outreach with lawmakers to achieve legislative changes and insurance reimbursement for NIPT. For example, in 2019 CAPS held a policy briefing at the Washington State Capitol where they recruited state lawmakers

technology (as for example are 23andMe or AncestryDNA genetic tests), genomics companies including Illumina, Invitae, Myriad Women's Health, Labcorp, Integrated Genetics, and Natera intensively marketed the technology to patients and providers when it was first released, encouraging patients to discuss this testing option with their reproductive physicians and physicians to offer the screening to patients. Indeed, NIPT was initially marketed to expecting parents as 'the gender test,' for its ability to identify fetal sex chromosomes, which continues to play in a big role in why pregnant people pursue this test. These companies' messaging focuses on "empowering informed choices," (Illumina 2023) urging expecting parents to "take control of (their) health with the power of genetic insights" (Myriad Women's Health 2023). Highlighting that "1 in 300 pregnancies are affected by a condition, and that "1 in 800 births" are affected with Down Syndrome while "1 in 1,000 births" has an open neural tube defect (Myriad Women's Health 2023), genomics companies underscore that all pregnancies *could be* at risk and that NIPT would enable life-changing decisions around family health and reproduction.[7] Above all, their marketing reassures parents with more *control* over family-building ("You're in control" (Invitae n.d.)) and *earlier* insights into their to-be child's health needs (Illumina 2023; Integrated Genetics 2020).

These marketing strategies appear to have been rather successful in terms of the sizeable commercial market and investments into NIPT. In 2015 alone, Natera raised $200 million to further develop the NIPT technology. Illumina, arguably the biggest commercial player in NIPT, spent $506 million (16%) of its revenue in 2022 on research and development, which included 'VeriSeq NIPT Solution 2' – the 'new and improved' NIPT which utilizes the latest next generation sequencing (NGS) technology and offers comprehensive screening for a wider range of fetal genetic conditions. In years past, Illumina has acquired several companies that develop and commercialize NIPT, including Verinata Health for $300 million in 2012 and Nextera for $130 million in 2014 (Illumina 2023; Illumina Inc. 2023; Research and Markets 2021). The technology also occupies a striking and substantial global commercial market. In 2020, it was estimated that 1.5 million NIPT tests were performed worldwide, with that figure doubling in two years – to 3 million tests – by 2022 (Research and Markets 2021). Further, the global NIPT market in 2022 was valued at $3.8 billion, with the United States occupying by far the largest share of $2.30 billion (Grand View Research 2022). This global market is expected to grow significantly, reaching $10.2 billion by 2027 (or $13.1 billion by some estimates). Expectedly, the United States is poised to continue leading this market (Ravitsky et al. 2021; Research and Markets 2021). (Grand View Research 2022; Markets and Markets 2022)

However, despite the standardization of NIPT across prenatal medical practices and public health measures, its sizeable market share, and the relentless growth of genomics companies developing these technologies, there remains little to no regulation of how these tools are implemented (National Council on Disability 2019; Pergament and Ilijic 2014; Ravitsky et al. 2021).[8] This

and staff to attend various panels hosted by genomics industry stakeholders. Although these lobbying efforts fundamentally benefit these companies' profits, their efforts are often framed as reducing healthcare access disparities, with the Washing Capitol presentation titled "Access and Disparity Challenges for Noninvasive Prenatal Screening" (National Council on Disability 2019).

[7] These figures may be different based on the NIPT panel used and will likely change in future as more chromosomal and single gene conditions are added to NIPT panels.

regulatory vacuum around prenatal genetic testing leaves much of the guidelines and ethics around implementing the technology to genomics companies, professional medical associations, and individual healthcare providers. As a result, there is also an increased expectation on individual patients to independently learn about NIPT and similar technologies, to use them for 'responsible reproduction.' In this way, the longstanding (and once State-run) goal within the field of human genetics to guide reproduction away from "less favorable" endowments can be gradually transmogrified into privatized 'liberal eugenics' wherein one's reproductive and testing choices are framed as matter of 'procreative autonomy' but are indeed undergirded by systemic social pressures and technological imperatives (Fox 2012, 2018; Mills 2013).

The Experts

Reproductive genetic technologies have led to new professions and experts. Of these, genetic counselors, geneticists, and reproductive physicians have been particularly relevant when it comes to implementing prenatal genetic testing for patients. The United States' trajectory of genetic counseling follows a similar timeline as that of developments in prenatal genetic testing. While early heredity clinics often performed genetic counseling (and coined the term, as discussed), the first official genetic master's counseling program emerged in 1969 at Sarah Lawrence College, at the same time that amniocentesis was taking hold in clinical settings. Several others, including at the University of California Berkeley, soon followed suit in the 1970s, further institutionalizing genetic counselors as key professionals in this arena (Minna Stern 2012:5, 17). These programs targeted middle-class, white mothers, presenting genetic counseling as a 'respectable' option for women to participate in the workforce. Moreover, the profession allowed for flexible schedules, where women could still fulfill their primary duties in the household. These demographic trends in genetic counseling have remained steady over the past decades, with 95% of counselors identifying as women and 90% as white or Caucasian in 2019 (Minna Stern 2012:3; National Council on Disability 2019).

Today, becoming a genetic counselor in the United States requires a two-year master's program (Minna Stern 2012). The counselor's main job is to "reliably translate (genetic) test results and technological language for a diverse clientele, aiming to equip them with equal doses of scientific acumen and human empathy to make decisions about their options" (Minna Stern 2012:1). With regard to prenatal genetic counseling, counselors' support parents in making "autonomous decisions about screening, diagnostic testing, possible preparation for the birth of a child with disabilities, or pregnancy termination" (Minna Stern 2012:15). Counselors play a central role in shaping how patients and physicians interpret prenatal genetic tests and approach subsequent decision-making. In recent years, reproductive medicine has seen an influx of genetic counselors needed int this space, with almost 50% of all genetic counselors practicing in reproductive medicine subfields (National Society of Genetic Counselors 2019). Still, the National Council on Disability continues to underscore that genetic counselors need greater disability education during

[8] Marketed as a 'laboratory-developed test,' NIPT's regulation falls largely to the Centers for Medicare and Medicaid Services (CMS) under the Clinical Laboratory Improvement Amendments of 1988. While CMS regulates laboratories performing genetic tests for analytical validity standards, there is no robust regulation over NIPT's use in clinics and around patient engagement. The Federal Trade Commission (FTC) does not specifically monitor genomics companies' messaging around prenatal genetic testing, and, perhaps most notably, NIPT is not approved or regulated by the Food and Drug Commission (FDA). In this context, it is imperative that research continues to study how NIPT, and prenatal genetic testing more generally, are unfolding and being routinized, highlighting opportunities for greater equity in how these technologies are used. (National Council on Disability 2019; Pergament and Ilijic 2014; Ravitsky et al. 2021)

their master's programs, in order to provide more balanced support for families whose pregnancies and children have been diagnosed with genetic conditions and disabilities (as opposed to the largely biased, negative information patients receive about raising a child with disabilities) (Meredith et al. 2023; National Council on Disability 2019).

Geneticists also play a central role in prenatal genetic testing, both in laboratory and medical settings, and for research and clinical outcomes. Geneticists are medically trained as doctors and can work closely with patients and other providers (including counselors and other physicians) on in-depth genetics needs. Many are also situated in academic settings, where they perform scientific research to better understand genetic markers of disease and conditions and inform technological development and diagnostic processes. ObGyns and Maternal Fetal Medicine (MFM) specialists – doctors who are specifically trained to manage high-risk pregnancies including those with genetic conditions – are also physicians involved in implementing prenatal genetic testing on a clinical front, but their roles are not focused on genetics and related patient engagement. As physicians focused on genomics, medical geneticists are critical to prenatal genetic testing, including informing the development and implementation of tests and ensuring patientcare in terms of accurate testing and results interpretation.

The rapid uptake and reliance on genetic testing are reflected in the demand for genetic counselors and medical geneticists. In their 2019 workforce report, The National Society of Genetic Counselors (NSGC) estimates that there is 1 genetics professional for every 300,000 patients, suggesting a dire shortage of genetic counselors in the United States (National Society of Genetic Counselors 2019). The NSGC projects the demand for genetic counselors will continue to grow by 28% from 2020 to 2028, based on how genetic testing technologies are being routinized across medical specialties (the Bureau of Labor provides a similar outlook) (Bureau of Labor Statistics 2022; Healthcare Management 2023). Similarly, the Bureau of Labor Statistics estimates that the employment of geneticists will also grow by 27% from 2020 to 2030, much faster than the average for all other occupations (Bureau of Labor Statistics 2023). For comparison, other healthcare roles are projected to grow 14% during this time, with counselors and geneticists representing almost double that rate (Bureau of Labor Statistics 2022, 2023; National Society of Genetic Counselors 2019, 2021a). Accordingly, there is a heightening demand for genetics experts who can appropriately implement genetic testing and address the needs of the precision medicine era.

This growth in prenatal testing and demand for these experts is marked by tensions in how reproductive genetic tools should be implemented. Geneticists, genetic counselors, and reproductive physicians are often trying to keep up with the pace of technological innovation when understanding how best to offer these tools to patients; this can lead to key jurisdictional tensions among these professional groups. New prenatal genetic technologies are developed– and marketed intensively by genomics companies – quicker that these providers can establish the best practices around how and when to implement them. As providers and patients work on-the-ground to use these technologies while they are still being developed and understood, there are numerous contentions and challenging consequences that emerge among them.

Emerging Issues in Prenatal Genetic Testing

Historically, genetic testing has been used as a tool for selecting against disability and promoting narrow ideas of 'normative' embodied ability. These eugenic ghosts continue to root many of the

persisting problems in reproductive genetic testing. As prenatal genetic testing has been marketed to patients and providers with little regulation, and providers continually play 'catch-up' against the pace of commercial innovation, there are several challenges that result from uncritically routinizing these technologies without thoroughly understand their social implications (this is especially the case with NIPT, the 'easy' 'game changer' prenatal genetic test). When it comes to health and social inequities, issues emerge around novel tools like NIPT because there is lacking expert capacity and limited genetics providers compared to how widespread and core to public health administration these tests have become. Not only are there systematic issues around patients' uninformed consent (as NIPT often gets ordered by ObGyns as standard practice, rather than through pre-test counseling with a genetics provider), but these problems are exacerbated when NIPT returns unclear or incorrect results, or triggers further genetic tests, putting patients into a whirlwind of testing that they may not thoroughly understand, for which they may not have been prepared and counseled. (California Department of Public Health 2023; Minear et al. 2015; National Council on Disability 2019; Pergament and Ilijic 2014; Rapp 1994, 1998; Rayna Rapp 2001; Ravitsky et al. 2021)

In what follows, I explore three key challenges with prenatal genetic testing, which inform the focus of this dissertation. First, I discuss the moralized making of the 'good mother' – the gendered social pressures on individuals (often times, women) to pursue prenatal genetic testing as a matter of reproducing responsibly. (Denbow 2015, 2019; Ikemoto 1992; Rapp 1994, 1998; Waggoner 2017). Here, we see how the technological imperative to prenatally test is encased in socioeconomic values of neoliberalism, privatized health responsibility, and heightened medical surveillance (Brown 2015; Clarke 2008; Reuter 2007; Rose 2001, 2008). Next, I delve into how disability de-selection is baked into systemic provider biases as they routinely use genetic testing for pregnancy management. Here, I highlight the relationship of prenatal genetic testing tools to the framing of disability, as reproductive providers and medical institutions (often implicitly) frame disability as an unwanted, tragic event, in their reliance on prenatal genetic testing (Meredith et al. 2023; Minear et al. 2015). Finally, I delve into the changing conceptions of healthiness and disability alongside genetic technological innovations, revealing how disabled communities have been steadily impacted by the standardization of prenatal genetic testing. Prenatal genetic testing has had striking consequences on reproductive decision-making patterns, diminishing social visibility and supports for those who have certain disabilities and conditions (Galis 2011; Norton et al. 2015; Zhang 2020). Here, we see how ideas around who represents 'meaningful existence' are narrowed as genetic tests increasingly provide insight into more genetic variations and frame these differences as pathological. Together, extant research raises questions around the social inequities and eugenics ghosts that remain, as routinized prenatal genetic testing emphasizes genetic determination and individualized approaches to health management, rather than holding social institutions accountable for nurturing inclusive structures.[9]

[9] A main concern around genomic technologies pertains to 'genetic determinism' – the idea that one's health and social outcomes can be essentialized to their genes. In her foundational piece, Abby Lippman discusses that, as more conditions (e.g., cancer, alcoholism, schizophrenia) are reconstructed as genetic diseases, patients may be at a disadvantage if genetic determinism precludes a structural or sociopolitical analysis of their health and related remedies (Lippman 1992). There are also concerns about how routinizing genetic testing may engender an overly individualized approach to medicine, one that places unreasonable accountability on patients to navigate personalized healthcare based on their genetic markers, neglecting broader gene-environment contexts (Rose 2001, 2008; Shostak 2003; Timmermans and Kaufman 2020). This is particularly salient in prenatal genetic testing, where pregnant people can be socially pressured into genetically testing, at times terminating, fetuses based on expectations to manage their

Testing and the "Good Mother"

Women's bodies have long been a key site for defining and regulating biopolitical citizenship through medicalizing reproduction. Women have been held morally responsible for 'reproducing judiciously' and ensuring children's' health (even before they are pregnant), while their reproductive autonomy has been continually constrained by legal and political systems (Armstrong 1998; Center for Reproductive Rights 2022; Denbow 2019; Waggoner 2017). Further, while the reproduction of white, class-privileged, able-bodied women has been celebrated, that of those outside this category has often been condemned (Roberts 1993, 2012). In the medical sphere, Black women's care and needs have been willfully neglected (Centers for Disease Control and Prevention 2023). Gynecology, in particular, was founded on experiments and un-anesthetized surgeries conducted on enslaved Black women considered to be 'sub-human' (Bailey and Peoples 2017). On the legal front, courts have sided with physicians to violently and forcibly discourage certain women's reproduction and override their prenatal decisions, supposedly in the interest of 'good mothering' and protecting the family institution (Bailey and Peoples 2017; Ikemoto 1992). Throughout the United States, and especially in California, there are well-documented cases of systemic sterilization and criminalization of parents of color and those with disabilities (Apple 1995, 2014; Ikemoto 1992; Manian 2020). Women who are marginalized, including sex workers or those addicted to substances, have been criminalized for not fulfilling their "responsibility of maintaining and transmitting family" that aligns with traditional values (Ikemoto 1992:6, 9). As such, defining citizenship along biopolitical lines has long depended on unequal "color-coded, class-coded, and culture-coded" understandings of motherhood, and parenthood at large (Denbow 2015, 2019; Ikemoto 1992:2-5,11).

This regulation of women's reproductive bodies and the pressures on individuals to mitigate health risks (and preclude reproduction of non-normative existence) are especially amplified within a privatized neoliberal landscape such as the United States. Scholars have defined the neoliberal context as one wherein each aspect of human existence is justified with reference to a "rational" framework of economic productivity (Brown 2015). Others have applied a neoliberal analysis to defining our current 'biomedicalized' medical setting, where medicine stands as a private industry, risk surveillance and technological reliance are heightened, and patients are expected to be informed, rational partners in privately managing their health (Clarke et al. 2003). With regard to prenatal genetic technologies, a rational actor in the neoliberal biomedical era is encouraged to act responsibly with regard to using these tests (also discussed as a 'technological imperative'), comporting themselves in a manner that "meshes with the morality of the state and the health of the economy" (Brown 2015; Denbow 2015, 2019; Waggoner 2017). Historically, we see prenatal genetic testing positioned as one's social obligation, as the imperative to test is framed alongside ill-health and disabilities being considered a family and public health burden. Some of the earliest justifications urging pregnant people to test and terminate fetuses with Down syndrome and trisomy 18 touted a cost-benefit analysis of saving $66,000 and $38,000 for males and females, respectively, illustrating larger sociopolitical pressures to de-select disability and reproduce

families' health (Minear et al. 2015; Rapp 1994, 1998). As such, researchers problematize how increased reliance on genetic technologies, without appropriate contextualization, furthers an understanding of health solely based on genetic constitution and reinforces historical inequities among diverse patient groups (Timmermans and Kaufman 2020).

'responsibly' as a matter of rationality (Minna Stern 2012; Waggoner 2017). Today, prenatal and newborn screening programs (including those incorporated into State public health infrastructure, as in California), as well as recommendations for all women of reproductive ages to diligently manage their reproductive responsibilities, continue to be justified using cost efficiency, framing caring for those who are disabled as a private challenge and socioeconomic burden that pregnant people should avoid (Meredith et al. 2023; Timmermans and Buchbinder 2012; Waggoner 2017).

The reliance on genetic technologies takes center stage when compelling pregnant people to individually ensure the health of their fetus and judiciously reproduce. In neoliberal settings, the responsibility for managing health risk is privatized and shifted onto pregnant people and individual families (Armstrong 1998; Clarke et al. 2003; Rose 2001, 2008). Individuals, particularly pregnant women, are spotlighted in terms of reducing the 'burden' of disability for the State and reproducing 'healthy' societies (Denbow 2015, 2019; Lippman 1992; Pergament 2013; Roberts 1993, 2012; Waggoner 2017). This is not to say that pregnant patients do not opt into prenatal testing; these technologies have been incredibly useful for those who seek genetic health insights. Many pregnant individuals look to prenatal genetic testing for ressurance of a 'normal' 'healthy' pregnancy ("relief from uncertainty"), or to terminate an unwanted pregnancy (Pergament 2013; Pivetti and Melotti 2013:77–78; White 1999:14). On the other hand, studies also find that people reject prenatal testing if they have doubts about risks posed by testing, including miscarraiges, result reliability, and potential emotional and psychological repercussions. Others also decline testing or pregnancy interventions based on how they discrepantly define disability and the need for 'normative' existence. The denial of testing can be a contentious moment between providers and patients (Piepmeier 2013, 2015; Pivetti and Melotti 2013; Rapp 1994, 1998). Those who choose to forgo use of their reproductive provider's medical guidance, do not use prenatal technologies, or who continue a pregnancy with knowledge of fetal disability, are often framed as pathological, irrational, or irresponsible (Blume, Galis, and Pineda 2014; Brown 2015; Denbow 2015; Lalvani 2016). These frameworks around risk, responsibility, and reproduction thus frame being a 'good mother' as one who uses prenatal technologies to ensure the genetic health and able-bodied-ness of their fetus, with substantial social pressures compounding this routine expectation to test one's pregnancy and select against disability (Apple 1995, 2014; National Council on Disability 2019; Pergament 2013; Roberts 1993, 2012; Waggoner 2017)

The Ghost of Eugenics in the Medical Institution

Among the social pressures on patients to pursue prenatal genetic testing are providers' biases and medical institutional patterns standardizing the use of these tools as a 'solution' to precluding births of children with disabilities and genetic conditions. There are instances where patients may not understand that they are undergoing prenatal genetic testing, or they may feel pressured to agree to testing because a provider has recommended it. The power imbued in a physician's medically authoritative role is significant, and when providers are biased toward testing (and against disabilities) this has an undeniable influence on pregnant patients and their families. Pressures on patients to prenatally test have been documented before the rise of NIPT, surrounding other prenatal genetic tests, but have been found to be exacerbated as NIPT has taken hold in recent years. As NIPT has emerged as an 'easily' administered test recommended for all pregnant people, ObGyns today often order this prenatal genetic testing without thoroughly counseling patients nor referring them for appropriate pre-test genetic counseling. As such, persisting provider biases lead to substantial concerns around lack of informed consent and systematic de-selection of pregnancies

with genetic variations, especially as many reproductive providers remain unclear on how to use prenatal genetic testing and make actionable sense of these insights (Bayefsky and Berkman 2022; Bernhardt et al. 1998). Without adequate genetics expertise and balanced counseling around disability, there is both an information vacuum as well as obscuring of patients' autonomy.[10]

Overall, studies show that when discussing prenatal genetic testing, ObGyns more negatively characterize the experience of raising a disabled child, even though research has long clarified that families with disabled children are no less fulfilled (Biesecker 1998; Parens and Asch 2003; Piepmeier 2013, 2015). A 2008 study found that only 29% of ObGyns "provided educational materials to their patients following Down syndrome diagnostic testing," while other studies note that physicians are less likely (compared to other reproductive health providers) to present prenatal testing as voluntary, instead directly recommending testing (Bernhardt et al. 1998; Meredith et al. 2023; Minear et al. 2015). More recent studies continue to find that uncritical routinization of prenatal genetic testing poses equity concerns for patients and dismisses their reproductive values. For instance, a 2017 Hastings Center Report found that providers discrepantly offered prenatal genetic testing to women based on their demographics, and differently supported their desires around continuing and terminating pregnancies. This report also problematized how the routinization of prenatal tests compromised patients' 'choice' around using these technologies and the reproductive decisions that follow: "many screening tests have been routinized in such a way that some women do not even recall agreeing to testing, while others feel that agreeing to testing is what their clinicians expect of them or that the testing is necessary to protect themselves and their families from the significant financial hardship of raising a child with a disability (Johnston and Zacharias 2017:1)." These concerns are especially salient for patients of low socioeconomic status with lower health literacy levels, who may not be able advocate for themselves and their reproductive priorities.

The largest study to date of parents who received a prenatal diagnosis of Down Syndrome (a condition with a relatively positive prognosis today), conducted between 2016-2021, found that physicians continue to present implicit and explicit biases when discussing prenatal genetic testing, framing disabilities as tragic and unwanted outcomes. Fewer than 40% of ObGyns discussed supports, services, and life outcomes surrounding Down Syndrome with patients, and over 60% presented the genetic finding as bad news or an undesirable testing outcome. Of these 60% of clinicians, only 17% provided patients accurate, updated resources about disability education (and

[10] Providers often situate their approach around prenatal genetic testing in empowering reproductive rights – that is, recommending that patients test their pregnancies with termination of disabled fetus' as a perceived advantage to doing so (Denbow 2015, 2019; Lalvani 2016). And indeed, historically, reproductive rights have been critical for pregnant people – enabling their ability to choose the reproductive outcomes concerning their bodies and fetuses. However, more recently, scholars have argued that this rights-based framework is limited and prejudiced against those with disabilities. The rights-based framework has been largely mobilized to emphasizes the choice to terminate pregnancies in cases where fetal health is compromised, but there is no similar celebration of the choice to bring to term a disabled fetus. It also echoes a long history of undermining and vilifying the reproduction of disabled parents, or those from other marginalized backgrounds (Piepmeier 2013; Waxman 1994). In response, Black feminist disability discourses have put forward the reproductive justice framework. Reproductive justice situates reproductive empowerment as a matter of social justice, contextualizes the individual as operating within a larger social structure, and celebrates more than the choice to abort or access contraception (Bailey and Peoples 2017; Denbow 2015; Piepmeier 2013, 2015). The justice framework fundamentally alters how we can conceive of disability and non-normal health, as it calls for a more equitable approach to enabling pregnancy, birth, and termination.

only half of the remaining 40% of clinicians, who did not initially present Down Syndrome as bad news, did so). Many parents described their physicians telling them "[you] could still miscarry, nature could still correct its mistake" or asking them prejudiced questions like "how does it make you feel to know that you will die and leave your baby dependent on the state?" and "how do you feel about your child not having good quality of life?" As this study was conducted well into the precision medicine and NIPT era in reproduction, we see how physicians (even those who are deemed to be unbiased in how they present findings of disability) are ill-equipped to discuss prenatal genetic testing in a balanced, equitable manner. This leads to serious concerns around the systemic use of these tools, as entrenched within medical institutions and their expert capacities, toward influencing patients' reproductive choices (even implicitly) and choosing against fetus' with disabilities. (Meredith et al. 2023)

Prenatal Genetic Testing and "Meaningful Existence"

Put together, the scholarship on how prenatal genetic testing is systematically used to reproduce able-bodied citizens suggests that these technologies have the potential to change how we consider healthiness and disability, and what it means to reproduce 'meaningful' existence. Fundamentally, research on the increasingly routine use of prenatal genetic testing shows how genetic insights are being marshalled to decide who is brought to life and who is 'de-selected' – with a gradual waning of certain disabled communities and the social supports that enable them to thrive (Piepmeier 2015, 2015; Rapp 1998; Rayna Rapp 2001; R. Rapp 2001). Consider Denmark, for example, which routinized prenatal genetic testing in 2004. In Denmark, almost all pregnant people undergo genetic tests and over 95% abort fetuses that indicate trisomy 21, despite comprehensive social supports for disabilities. As a result, in 2019 (15 years after prenatal testing was routinized), there were only 18 children born with Down syndrome in the entire country – the near disappearance of a disabled population (Zhang 2020).

The United States has followed along similar lines when it comes to prenatal testing and selecting against disabilities, with 70% of parents terminating pregnancies with Down's Syndrome between 1995 and 2011. Research suggests that there are around 6000 (by some estimates 5300) children born with trisomy 21 each year in the United States (National Council on Disability 2019; Natoli et al. 2012). And these trends hold for conditions that are rarer than Down Syndrome. An early study following prenatal carrier testing for Gaucher disease[11] found that of 26 affected pregnancies, 5 parents elected to terminate (20% terminated), even though "none of the 21 children homozygous for this gene mutation presented with severe disease after 15 years (Pergament and Pergament 2012:521–22)." Importantly, these studies do not represent the effect that the more recent routinization of NIPT in the United States has had on biases against disability. This is particularly relevant, as recent prenatal genetic innovations such as NIPT tend to identify more "life-limiting" than "life-threatening" disabilities compared to technologies past, positioning these are one in the same when it comes to identifying genetic variations and avoiding any such condition (Pergament

[11] Gaucher disease is a rare genetic condition. Individuals with Gaucher disease are missing an enzyme used to break down lipids, impacting one's spleen and liver. While type 2 Gaucher disease may lead to early death, those with type 1 have normal life expectancy with enzyme replacement therapy. (Mount Sinai 2023)

2013).[12,13] Still, extant scholarship focused on how biased implementation of prenatal testing impacts those with disabilities highlights the harmful consequences for social equity, particularly the stigma and other socioeconomic costs that those who forgo testing or live with disabilities increasingly face. (National Council on Disability 2019)

Unlike places like Denmark, the United States presents a context where the management of health and disability is largely privatized to individuals, which has notable consequences on the social perception (and inclusion) of disability. In this setting, disability is framed as biologically determined and as a private burden; with lacking public disability supports, it can be unfeasible for families to use their resources toward supporting a child's additional needs (Denbow 2015, 2019; Waggoner 2017). Where problems are attributed to one's embodied disability, research shows that many such challenges are rooted in insufficient and unequitable social arrangements that indeed can be changed and restructured if appropriate resources are redirected to doing so. Instead, what we see in the United States is an emphasis on using prenatal genetic testing to preclude disabled existence because disabled individuals are societally positioned as having inherently less fulfilling lives and less capable of fruitful social participation (Asch 1999; Garland-Thomson 2015; Parens and Asch 2003; Pergament 2013; Pergament and Pergament 2012). Further, in the privatized health landscape of the United States, commercial marketing compound biases against disability; commercial tactics often message genetic testing as more urgent, necessary, and deterministic than the probabilistic risk-score results these technologies are able to offer (Biesecker 1998). This contorted framing of prenatal risk leverages patients' vulnerabilities, which during pregnancy decision-making can have severe outcomes. Thus, where disability is a private 'burden,' the gap around disability social supports and inadequate social education regarding disability resources can make it challenging for expecting parents to consider their full range of reproductive choices in the face of prenatal genetic findings. (Asch 1999; Minear et al. 2015; National Council on Disability 2019; Natoli et al. 2012; Parens and Asch 2003; Piepmeier 2013, 2015)

With more insight into prenatal genetic health, and technologies that identify (and medicalize) even mild genetic variations, we can perhaps expect more stigma and selection against fetuses with disabilities and genetic conditions. As the following chapters illuminate, the routine and uncritical use of these technologies have a gradual yet undeniable effect on positioning disability and genetic difference as unwanted, systematically threatening the existence of disabled communities and those with genetic variations. I underscore that this critique of how prenatal genetic technologies have been implemented is not to undermine one's reproductive choice – individuals should be able to safely terminate pregnancies for whatever reason they choose, even if that choice hinges on their perception of disability. As the above scholarship suggests, and as this dissertation will reveal, the issues around deselection of disability emerge when these patterns are systematized, baked into provider biases, institutional practices, commercial marketing, and broader social pressures and socioeconomic contexts that are fundamentally intolerant of diverse existences.

[12] More recent data on termination patterns around disability is still underway, as newer technologies like NIPT were released in 2011 and steadily routinized in the years following (with the ACOG recommendation to offer NIPT to all pregnant women coming in 2020) (National Council on Disability 2019).

[13] For many 'limiting' conditions, a person may require additional supports to facilitate their wellbeing and social participation, but their condition would not jeopardize their life overall. Life 'threatening,' conditions, on the other hand, are typically fatal. (Pergament 2013)

Questions

The chapters in this dissertation explore several critical questions that are grounded in the extant social problems and inequities around prenatal genetic testing as outlined above. Overall, I ask, how does the "appropriate" use of prenatal genetic technologies come to frame certain existences as meaningful and worthwhile while others are seen as unwanted and less valuable? In doing so, I also question the role that experts play in both implementing and interpreting these technologies during medical research as well as patient care. Further, I examine the implications this reliance on prenatal testing has for those who are pregnant in terms of their responsibility toward managing family health.

The first chapter tackles each of these three overarching questions by examining the adjudication of disputes between patients and providers regarding prenatal genetic testing and the birth of disabled children. We see how the use of prenatal genetic tests and resulting 'injuries' of disability have been defined and deciphered within courts over the past six decades. As courts offer judgements on Wrongful Birth and Life cases, how do they position disability as a legal harm? And how do they frame the expectations to use these tools and the responsibility (of parents and expert providers) to prevent these so-called harms? As the technological imperative around prenatal genetic testing emerges, there are consequences for how we understand disability and the role of experts and individual parents in preventing these outcomes.

The second chapter dives deeper into the question around how prenatal genetic technologies contribute to changing conceptions of disability, meaningful existence, and what it means to embody health. Specifically, I explore how prenatal genetic tools are used socially construct diagnostic categories that influence reproductive decision-making. How do emerging diagnostic categories based on genetic differences cast certain existences as normal while others are seen as medically abnormal and perverse? I examine these issues through a focus on sex chromosome aneuploidies, a growing and rapidly changing prenatal genetic diagnostic category. In addition to the biases in evolving prenatal medical pathology, this chapter also explores the questions around consequences for those who are pregnant or living with genetic variations and disabilities.

Finally, the third chapter centers on the question of experts. I examine the types of experts and expertise that are necessary and meaningful for the current era of precision medicine in reproduction, uncovering current challenges and tensions among professional groups. As new prenatal genetic technologies are routinized, there are changing ideas around which professionals, with their distinct skillsets, are needed to equitably operate the space and meet patientcare demands. How do various groups of experts shape the current medical moment in prenatal genetic testing, and in doing so, how do they define the social values around implementing prenatal genetic technologies? Here, I focus on the dynamics and perspectives among reproductive physicians and genetic counselors working with prenatal genetic testing.

Methods

I use mixed qualitative methods to address the questions discussed above. First, I conducted 60 in-depth interviews (20 with reproductive physicians, 20 with genetic counselors practicing in prenatal medicine, and 20 with patients who have used or declined prenatal genetic testing). Second, I completed ethnographic observations at various sites: professional genomics conferences attended by geneticists, reproductive physicians, genetic counselors, and patient

communities around the United States; a genetic counseling master's program; and consultations between patients and genetic counselors at a large genetic counseling clinic in California. Third, I conducted content analysis, using a Python-assisted web scraper to gather text-based exchanges from prenatal genetic testing related threads on Reddit, a popular online discussion forum. Finally, I conducted archival research and analysis on Wrongful Birth and Life cases that pertain to the use of prenatal genetic testing technologies in the United States over the past almost six decades.

California: When recruiting interviewees, I sampled patients and providers who worked with prenatal genetic testing in California. Additionally, archival research focused on California cases, although influential cases from other states were also included. I also observed patient-counselor consultations that took place in a California clinic. The state of California was particularly relevant because the routinization of genetic testing during reproduction is particularly visible in this state, which has long been a "hub" for assisted reproductive services including prenatal genetic testing (Dunn 2019). California was an early adopter of widespread prenatal genetic testing, with the 1986 California Screening Program that pledged state-wide access to maternal serum alpha-fetoprotein (MSAFP) testing. As discussed, today, the California Prenatal Screening Program covers almost all major prenatal genetic tests, including the more recent NIPT, allowing wide implementation of these technologies across medical institutions and patients of varying backgrounds (California Department of Public Health 2023). California also contends with a fraught history of eugenics as it stands at the cutting edge of biotechnology and genetics. The state has conducted perhaps the most systemic reproductive abuse in the United States' recent history, leading the country's eugenics agenda since the 1930s. Between 2006 and 2012, over 150 incarcerated women (mainly of color) were sterilized in California prisons, which provides insights into how newer genetic technologies may be managed against this looming historical background (Denbow 2015:141; Kaelber 2012; Manian 2020; Watanabe 2020). Finally, California is also a budding ground for new genetic counseling programs that are remerging and redefining their practices against eugenics legacies, including at University of California Los Angeles, University of California San Francisco, and Stanford University. Per the 2019 National Society of Genetic Counselors report, California is also home to the highest proportion of genetic counselors nationwide, and third highest according to the Bureau of Labor Statistics (Bureau of Labor Statistics 2022; National Society of Genetic Counselors 2019). Thus, California represents a setting where reproductive medicine experts are consistently engaging with genetic testing, and the way these innovations impact their relative expertise, workflow alongside other experts, and approaches to patient care. Patients and providers in California contend with considerations at the forefront of genetic testing, which are not as readily realized in other places. Thus, the perspectives and narratives from those engaging in California's prenatal genetic testing landscape are particularly influential when understanding how these technologies are changing reproductive experiences and ideas around health and disability.

In-depth Interviews. I conducted semi-structured in-depth interviews with California-based reproductive physicians (ObGyns and MFMs) (20), genetic counselors (20), and patients/individuals (20) who have used (or declined) prenatal genetic testing. Each interviewee category had a customized interview protocol depending on their role in prenatal genetic testing; however, protocol themes overall paralleled each other, allowing a comparison of each group's perspectives on a shared set of issues. Protocols were informed by existing theories and empirical observations (Rapp 1994, 1998), as well as questions pertinent to the scope of this project.

Interviews revealed how each actor approaches prenatal genetic testing and how they considered their roles in relationship to one another (highlighting moments of collaboration and contention). They also illuminated thought processes around reproductive decision-making after prenatal testing, and how each group evaluates the use of prenatal genetic technologies in the reproductive process. Each interviewee discussed how they understand and negotiate health and disability as a result of prenatal testing, and their experiences when pursuing or forgoing testing, or interpreting complex results.

Interviewees were sampled using a variety of methods. First, using interval sampling methods, physicians and genetic counselors with their emails listed publicly in professional society directories (i.e., Society for Maternal-Fetal Medicine, National Society of Genetic Counselors, American Society for Reproductive Medicine) were contacted and invited to participate, if they were providing care in California. I also invited provider participation using public posts on online forums for these professionals, including the Twitter community frequented by genetic counselors. Finally, interviewees provided references to colleagues and helped circulate the call for participation within their professional networks, generating a snowball sample. The final sample of interviewees represented professionals working across a diversity of settings, from academic medical centers, genetics laboratories and private biotechnology companies, community and non-profit hospitals, private clinics, and large general hospitals. These various institutions were located across California, including in urban and rural areas, low-income communities, and affluent neighborhoods, ensuring that interviewees could illuminate a range of perspectives and experiences. Those interviewed were also at varying career stages, with counselors and doctors ranging from early practitioners, with 1-2 years of experience, to those with decades of experienced or recently retired. Patients also represented various ethnoracial backgrounds, income levels, and health literacy and education. They were recruited largely through social media posts on Reddit and Twitter as well as snowball sampling.

Ethnography. In order to understand how knowledge around prenatal testing and health is being produced and negotiated in provider communities, I observed distinct sites for information engagement. Given the constraints of the pandemic during the data collection period, all ethnographic observations were conducted digitally (via Zoom or conference hosting platforms). Where provider interviews gave insight into their self-reported perspectives and experiences of prenatal testing, the ethnographic observations of professional conferences and provider-patient consultations illuminated their knowledge production processes and more unrestricted expressions among colleagues of how prenatal genetic technologies should be developed and used. For both genetic counselors and doctors, I attended professional genetics conferences that had panels pertinent to prenatal genetic testing. To create a sample of conferences, I compiled a list of major genomics conferences between 2021 and early 2022 (I corroborated this list with feedback from providers regarding which conferences they usually attended) and prioritized those that I could attend virtually as a non-provider. I attended each conference as a virtual observer and took copious fields notes (and screen grabs of presentation slides), without engaging in the chat or discussion, with my video turned off, so as to minimize my presence and impact on the discussion among providers. [14]

[14] Ultimately, I attended 7 professional genetics conferences where I took detailed observations of each session relevant to prenatal genetic testing: Philadelphia Prenatal Genetic Diagnosis Annual Conference, Genetics Society of America "Reproductive Genetics" Conference, Society for Maternal Fetal Medicine Annual Conference, American

Interested in learning more about how genetic counselors approached their work, I observed prenatal genetic counseling consultations between genetic counselors and patients at a large hospital that served individuals of all income and ethnoracial backgrounds in California. Here, I was virtually present for each consultation; while each patient had to consent to my presence, I remained silent, with my video turned off, so as to minimize my presence during the consultation. I conducted a total of 60 hours of these prenatal genetic counseling observations. I also attended courses at genetic counseling master's programs. I observed a highly ranked genetic counseling master's program, sitting in on three courses where counselors-in-training debriefed their prenatal clinical cases.

Web-Content Analysis. I conducted a content analysis of patient community discussions regarding prenatal genetic testing and reproductive decision-making. Like the ethnography, this provided insight into knowledge production, health negotiation processes, and key logics within patient communities. I used Python to build a web scraper that gathered this content from Reddit based on keywords, such as: prenatal genetic testing, results, gene, baby, chromosome, terminate. To build the scraper, I manually identified sub-Reddits (discussion forums dedicated to a specific topic) pertaining to prenatal genetic testing. I narrowed the selection of sub-Reddits based on the number of relevant keywords present and the number of active users who engage with the sub-Reddit, providing a sample of 18 sub-Reddits where patients were actively discussing their perspectives and experiences with prenatal genetic testing. I then built a Python program to search each sub-Reddit for associated keywords and compile a document of all discussion threads which had one of more of those words (by default, the program pulled the top 7 threads from each keyword and sub-Reddit pairing). At first, the scraper provided 944 threads, which were cleaned (removed duplicates and irrelevant threads) to generate a final list of 233 discussion threads.[15] Reddit provided an optimal 'field' to observe patient discussions and knowledge making, because it is a free and widely accessed online platform dedicated to targeted forums and questions (ensuring content relevance). Reddit users tend to be quite candid in their responses and comments to one another, allowing for insightful analyses. Although, it is also important to note that relying on Reddit has its limitations; users represent a self-selective community who are not only presumably tech-savvy but are also actively interested in engaging in public forum conversations.

Archival Analysis. Finally, I conducted archival research into Wrongful Birth and Life cases pertaining to prenatal genetic technologies in the United States.[16] Using the online database "Casetext," I narrowed this sample of cases to California or federal appellate courts, since federal judgements are persuasive nationwide. Casetext is a comprehensive legal database that includes

College of Obstetricians and Gynecologists Annual Clinical and Scientific Meeting, American College of Medical Genetics and Genomics, National Society of Genetic Counselors Annual Conference, Southern California Genetic Counselors 4th Annual Education Conference. The sampling of conferences was not restricted to those in California, as knowledge production communities almost always extend further than one's geographic location of practice and experience.

[15] The cleaning and process of these Reddit discussion threads was greatly enabled by my undergraduate research assistants, Natalie Rivas, Karen Tirado, and Elizabeth Brown.

[16] This data was collected and analyzed alongside a co-researcher, Zaina Mahmoud. It also contributes to a separate law-focused article in-progress.

all federal and state cases and statutes above trial court level. The sample included all Wrongful Birth and Life cases heard at the various court levels in California, including the Supreme Court, Court of Appeals, and District Courts. It included judgements published between 1963 and 2021, and close analysis was undertaken for cases between 1963 and 2015. The sample also encompassed landmark and especially influential Wrongful Birth and Life cases from other states outside California. In total, 37 cases were analyzed, 16 of which were Californian cases. Of the California judgements, 9 were Wrongful Life claims, 2 were Wrongful Birth claims, and the remaining 5 were Wrongful Birth and Life claims. This archival research enabled a historical understanding of how prenatal genetic technologies have been developed and interpreted in the United States, marking significant transformations in ideas around how these technologies should be 'correctly' used and the implications these tools have for defining disability and meaningful existence.

Analysis. Each data collection method provided text-based data (transcripts, fieldnotes, scraped text content, legal cases) that was qualitatively coded using the software MaxQDA. I applied the same codebook to each dataset (with the exception of archival legal cases, discussed below), based on themes from existing literature as well as emergent findings, largely resembling abductive analysis (Timmermans and Tavory 2012). This maintained consistency in overarching themes and patterns being studied, enabling comparison across groups and datasets. The archival legal cases were analyzed using a separate codebook, which accounted for historical themes and over-time developments, as well as functions specific to legal judgements (i.e., damages, charges). However, the results from the legal case analysis closely parallel themes in the rest of the datasets, allowing for a comprehensive project around prenatal genetic testing and its impacts on health and disability, reproductive decision-making, and implementation challenges among patients and providers.

Overview of Empirical Chapters

This dissertation explores the complex landscape of prenatal genetic testing, aiming to unravel the implications of its routine implementation in contemporary healthcare (that is, in today's precision medicine era). Across three empirical chapters, this research scrutinizes the evolving dynamics, social consequences, and shifting paradigms associated with prenatal genetic testing. Each chapter unravels a distinctive facet of how prenatal testing has shaped societal approaches to reproduction, health, and disability, shedding light on the far-reaching impact of prenatal genetic technologies on our society.

In the first empirical chapter, I delve into the realm of Wrongful Birth and Life cases pertaining to prenatal genetic testing disputes over the past six decades. The analysis illuminates courts' evolving interpretations of how prenatal genetic technologies "should" be used. The chapter reveals how technological advancements have not only expanded health and risk management expectations but have also introduced increased individualized responsibility, placed on both parents and healthcare providers. Disability, in this context, becomes framed as unwanted and an existence to be precluded through the use of prenatal genetic testing.

I focus on legal institutions because their judgments, in part, regulate our relationships, norms of moral conduct, and establish the stakes for partaking in society. Moreover, in evaluating their written judgements, we rather clearly observe shifts in social attitudes and approaches to broader issues — in this case, pertaining to the correlation between reproductive genetic technologies, privatized health responsibility, and meaningful existence.

The main takeaways from this chapter underscore the transformation of disability as an unwanted existence, alongside the interpretation and routinization of prenatal technological developments in neoliberal settings. It becomes evident that disability is perceived as a societal burden, and that the responsibility to manage the health and care of disabled individuals is shifted to private individuals. Moreover, we see how courts judgements come to systematically prefer a so-called normative embodiment of productive and functional existence. The uncritical and routine implementation of prenatal genetic technologies can lead to lasting social transformations, particularly around the deselection and devaluation of certain lives, making this chapter a key historically grounded analysis of the ethical, legal, and societal dimensions of prenatal genetic testing.

In the second empirical chapter, I unpack the social making of genetic diagnostic categories and their impact on prenatal genetic testing. Using the case of how sex chromosome aneuploidies are socially constructed as a diagnostic category, this chapter offers an in-depth examination of the pathologizing of gender and the medicalization of genetic variations and conditions – which, today, occur earlier in the prenatal process than ever before. It highlights how diagnostic categories with unclear medical implications can complicate and compromise reproductive decision-making and patientcare, all while reinforcing social biases as a part of 'objective' medical science.

The chapter underscores how technologies are routinely and uncritically used to identify and pathologize genetic differences, even those such as sex chromosome aneuploidies which are often mild and inconsequential to one's overall wellbeing. These processes of pathology are increasingly enabled by in-utero insights into genetic differences, narrowing perspectives of diverse existences and reflecting more social biases around what it means to embody "normalcy." The chapter emphasizes that not every genetic difference needs to be diagnosed as a medical or pathological condition, especially when it may have little to no health implications. Further, diagnostic categories are revealed as representations of medical power, reflecting social prejudices, rather than being fixed, immutable categories. The routine use of genetic technologies, without consideration for which differences need to be identified and medicalized in-utero, renders certain existences as pathological and "abnormal" while emphasizing the need to use prenatal genetic technologies to choose against (or 'fix') these outcomes.

The third and final empirical chapter of this dissertation centers on the experts and expertise within the field of prenatal genetic testing, particularly focusing on reproductive medicine physicians and genetic counselors. This chapter delves into the intricacies of the relationships between these groups and how they organize their expertise and responsibilities around prenatal genetic testing. It sheds light on the strategies employed by doctors to 'shuffle' (i.e., delegate) aspects of specialized genomics expertise to counselors, who 'strategically absorb' these responsibilities are core to their professional roles. This process of shuffling and absorbing highlights the increasing value of socioemotional care and the navigation of ambiguity in precision medicine, positioning the genetic counselor's role as central to delivering equitable patientcare in prenatal genetic testing.

The central points from this chapter revolve around the transformative impact of sociomedical contexts on the reorganization of expertise and experts within reproductive genetic testing. It examines how the current context, where routine reliance on genetic testing is increasing while the values around patient-centered care are sustained, gives significance to the expertise

shuffling/absorbing between doctors and counselors. In doing so, the chapter emphasizes implications on which expert perspectives are influencing the forefront of genomics and equitable care, suggesting that genetic counselors perhaps have more potential to define this landscape than ever before. This shift in how expertise is organized and valued is changing the very nature of what it means to provide equitable patient care in reproductive genetic testing, ultimately elevating the genetic counselor from an auxiliary to a more central healthcare provider in reproductive medicine. Throughout, the chapter also presents the possibility that a larger transformation is taking place in terms of the experts and expertise defining reproductive genetic medicine, as the historically feminized and excluded perspectives of genetic counselors are becoming increasingly important to meeting emerging patient demands and delivering equitable healthcare.

Overall, this dissertation highlights the pressing need for a more research to inform careful implementation of prenatal genetic technologies, with a particular emphasis on how Non-Invasive Prenatal Testing (NIPT) is unfolding. The routine integration of these technologies into public health measures demands a reconsideration of our current approach. Commercial genetics companies and medical institutions have played a significant role in shaping the utilization of these tools, often without adequate input from patient communities and disability advocates. Further, it is imperative that we build the capacity of experts in medical fields that rely on genetics, in order to harness the benefits that prenatal genetic testing and NIPT can offer in terms of health and risk management, family-building, and reproductive choices. The consequences these technologies can have on social and health equity, reproductive decision-making, as well as the meaningful inclusion of disability and diverse existences, are profound, which makes close and ongoing examination of their implications that much more crucial.

To be sure, prenatal genetic testing itself is not inherently harmful; rather, harms result from how we systematically employ these tools to treat disability as objectively unwanted, often neglecting broader social consequences. The path forward necessitates more emphasis on equitable and inclusive social infrastructures that align with the development of reproductive genetic technologies. We cannot underestimate the power of inclusive social structures, even as these technologies help us identify genetic differences and better manage health needs. As such, it is crucial to foster sociocultural attitudes and structures that promote of tolerance alongside providing widespread access to prenatal genetic testing. Healthcare approaches – on both the medical institutional and individual provider levels – need to steer away from encouraging testing solely to prevent certain births, and instead incorporating these tools into an approach that advances reproductive justice and disability inclusion.

There are significant consequences surrounding the relationship between genetic technologies, systematic de-selection of disability, reproductive pressures, and the enduring history of eugenics. As such, as prenatal genetic innovations become more precise, capable, and accessible, it is essential to implement them in ways that contribute to a more inclusive and just society, rather than perpetuating exclusive measures and social pressures that marginalize disability and reinforce inequities. In that vein, this dissertation is an endeavor to pave the way for a more equitable and compassionate future in the realm of prenatal genetic testing.

CHAPTER TWO

The Technological Making of Disability: How Legal Adjudication of Reproductive Genetic Technologies Creates "Unwanted" Existence

This chapter was co-researched with Zaina Mahmoud, Ph.D. It has been substantially developed for this dissertation.

Notions of heredity – the idea that traits and conditions can be passed down among biological ancestors – emerged as early as the 1900s in the United States (Black 2003; Minna Stern 2012). Between the 1940s and 1950s, scientists identified DNA as 'the code of life' ("The Structure" 2016). Breakthroughs in genetics, and specifically cytogenetics (i.e., testing tissue, blood, or bone marrow for chromosomal changes), tangibly introduced the concept of genetic inheritance into reproduction and family-building. These emerging understandings of genetic science enabled the first innovations in reproductive genetic technologies during the 1950s. Among these early technologies was amniocentesis, where a long needle is inserted through a pregnant person's abdomen, into their uterus, to sample amniotic fluid in order to sequence fetal DNA and diagnose fetal conditions ("Mayo" n.d.). Technologies like amniocentesis paved the way for today's non-invasive prenatal testing (NIPT), which uses a standard blood draw from a pregnant person to screen for the likelihood a fetus has genetic conditions including Down Syndrome, Edward Syndrome, Patau Syndrome, and various sex chromosome aneuploidies. Over the years, as reproductive genetic technologies have been developed into more sensitive and reliable tools, they have become central to prenatal clinical encounters (Norton et al. 2015). With knowledge about a fetus's genetic state, healthcare providers can guide patientcare, and potential parents can shape their expectations for a child or make reproductive decisions that align with their priorities (Denbow 2015). In these moments, prenatal genetic testing technologies are often intertwined with decisions about continuing or terminating pregnancies based on genetic conditions, sex preferences, or other congenital differences.

Developments in prenatal genetic technologies, and the ways parents and providers use these tools, reflect and reinforce broader perspectives on disability, normalcy, and deserving existence (Meredith et al. 2023). One way to observe these societal ideas, especially as they evolve over time, is to consider how discussions about relevant technological developments unfold within social institutions. In this chapter, I focus on legal institutions – specifically, courtrooms – as they adjudicate the 'proper' use of prenatal genetic technologies during reproduction. Courts are critical spaces where the relationship between biotechnologies and disabilities is interpreted (Lukes and Scull 2013). When prenatal genetic technologies are being developed and yet to be 'perfected' in terms of their reliability or best practice guidelines for implementation, there are plenty of contentious moments regarding how they should be used and toward what ends. Providers and patients can be uncertain about when it is appropriate to use prenatal genetic testing, or what level of risk or error is tolerable. They may be unsure about how to navigate clinical interactions around tests and results. Each of these concerns has bearings on questions of life and existence.

How should expecting parents use prenatal genetic testing to inform reproductive decisions, if at all? How might providers support parents in this process, or at the least ensure parents have access

to reliable genetic information when making consequential pregnancy decisions? In what ways do these genetic technologies cast certain existences as worth living, and others as 'abnormal' and unwanted? And, most importantly, what does this mean for those living with disabilities and the way disabled existence is framed in light of genetic technologies that can identify (even mild) differences and conditions in-utero?

These questions around how societal perspectives of disability and meaningful existence develop alongside prenatal genetic testing innovations are particularly salient in the United States, where public supports and accommodating structures for families with disabilities are severely lacking. Here, disabilities are framed as a problem to manage individually, affecting a 'minority group' (even though disabled people are estimated to constitute over a quarter of the United States' population), rather than a challenge for *any* individual desiring full social participation where exclusive structures undermine the collective quality of life (Bowen 2022). As a result, raising a child with disabilities and existing as a disabled adult are largely private issues in the United States; families and individuals often rely on their own resources and community networks to secure necessary supports and accessibility measures (Zola 2005; Bickenback and Cieza 2011). In instances pertaining to reproduction and genetic testing, parents of disabled children may be left with no recourse other than to turn to legal avenues to secure resources to provide for their child's additional needs. As such, in the United States, courtrooms stand out as a fitting (and under-explored) site to observe how emerging prenatal genetic technologies change how we navigate genetic conditions, disabilities, and questions around who we 'should' bring to life. (Fox 2019; Mor and Pikkel 2019; Shanley 2002; Vecera 2014)

Courts invoke the torts of wrongful birth and wrongful life (WBL) to adjudicate appropriate uses of prenatal genetic technologies and regulate how these tools render 'acceptable' bodies and reproductive choices. Wrongful life (WL) claims are brought by the disabled child, claiming their disabled life as an injury sustained due to their healthcare provider's negligence. In WL claims, the child must assert that it would have been preferable for them to not exist at all, compared to living in their current condition. Wrongful birth (WB) claims are brought by the parents of a disabled child. Parents claim that their provider acted negligently around prenatal genetic testing, resulting in a failure to diagnose or disclose their child's genetic anomaly in-utero. Here, the injury is a deprivation of reproductive choice; parents must assert that they would have terminated their pregnancy, had it not been for the provider's negligence. Importantly, in both WL and WB cases, the injury is fundamentally the existence of the disabled child – in the former, the harm is the existence of a disabled life while in the latter it is the loss of the choice to abort a disabled fetus. In WBL claims, courts reason with the 'proper' use of prenatal technologies as related to understandings of 'valued' existence and acceptable family-building practices. As they attempt to reflect and regulate societal views through WBL cases, legal institutions become important arbiters of whose life has been 'rightfully' created and how diverse existences are prioritized, valued, or regretted (Lukes and Scull 2013). (Hensel 2005; Fox 2019; Mor and Pikkel 2019)

Tracing how wrongful birth and life cases have been adjudicated over the past 58 years, this chapter explores how sociocultural narratives around disabilities and reproduction are dialectically shaped alongside steady innovation and use of prenatal genetic technologies. How have societal perspectives about babies born with genetic conditions, their families, and medical providers changed with growing access to prenatal genetic insights and the possibility of altering one's

reproductive choices accordingly? How does increasing use of prenatal genetic technologies illuminate shifting perspectives on what makes for meaningful existence and who should be responsible for reproducing children along these lines? To answer these questions, I analyze how courts co-construct engagement with prenatal genetic testing, as these technologies shift from being novel innovations to becoming routinely implemented and standardized within prenatal care.

Delving into WBL cases, from the first claim in 1963 until 2021, I illustrate how courts articulate expectations for the 'appropriate' use of emerging prenatal genetic technologies, and in doing so co-produce societal narratives around reproduction and disability. These judgements reflect and reinforce technoscientific zeitgeists, wherein prenatal genetic technologies are gradually seen as a 'remedy' in precluding unwanted (and supposedly unproductive) disabled existences (Lukes and Scull 2013; Denbow 2015; Rapp 1998; Meredith et al. 2023). Though courts underscore the value of all life, the reasoning behind their adjudication positions disability as inherently undesirable and promotes the expectation that genetic technologies should be used to reproduce typical able-bodiedness. While certain births are legitimized as representing 'worthwhile' existence, others are considered results of medical or technological 'failings' due to their genetic differences.

The chapter highlights three discourses that emerge in WBL judgements, which illuminate the evolving relationship between biotechnology and desirable existence in the United States. First, courts hinge expectations of health and risk tolerance on the *'promises' of prenatal technology.* The way health ought to be techno-scientifically managed, and the room for error when doing so, are contingent on the contemporary capacities reproductive genetic technologies have to offer; the more diseases that can be detected with greater precision, the higher the expectation to use these tools. Next, responding to developments in prenatal genetic testing and the routinization of these technologies within neoliberal contexts, courts *privatize responsibility* for managing family health onto parents. Parents are held accountable for individually providing for their children's needs and opting into testing to 'judiciously' birth 'normal' children. Providers, too, are held individually responsible for utilizing genetic testing tools appropriately to preclude the birth of disabled babies. Finally, while life in itself is seen as inherently valuable, courts frame some *disabled existence as 'defective' and 'objectively' unwanted.* In grappling with these issues, courts mobilize key changes in technological innovations, medical recommendations, and legislation and policies around reproductive autonomy and fetal existence. Ultimately, these judgements transform congenital disabilities into legally cognizable injuries, reinforcing the imperative to use prenatal technologies to prevent such birth outcomes.

Developments in reproductive genetic technologies have a steady yet undeniable effect on broader conceptions of disability and meaningful existence. The way these ideas around existence evolve represent the sociotechnical context in which they are borne, especially regarding how disability is seen as a private or public concern and how technologies are used to 'manage' varied existences (Galis 2011; Winance 2016; Rapp 1998; Meredith et al. 2023). In the United States, prenatal technological developments, particularly in the way they become routinized in clinical care, represent the processes of "biomedicalization," wherein medical technologies are increasingly translated and integrated into ordinary life practices, as a result transforming bodies and identities in "technoscientifically enmeshed ways" (Clarke et al. 2003, p.162-166). As pregnancy becomes biomedicalized (in large part due to increasing integration of prenatal genetic innovations), disabled bodies and diverse existences are transformed and situated within a broader neoliberal

context, where each aspect of human existence is justified with reference to a "rational" framework of economic productivity (Brown 2015; Denbow 2015; Tseris 2018). Biomedicalizing (or technologically entangling) reproduction in this neoliberal setting gives rise to new technological imperatives – in this case, the pressures to use prenatal genetic technologies toward creating 'healthy' and 'productive' lives (per ableist terms) (Rapp 1998; Meredith et al. 2023). Over time, such ideas around how testing innovations 'should' be used reinforce and continually shape societal values around inclusivity, disabled existences, and what represents a meaningful life. (Fox 2019; Mor and Pikkel 2019; Vecera 2014)

When disabilities are viewed within a neoliberal context, they are cast as inherently deleterious to an economically productive society and further entrenched as unwanted private burdens. The rapid and uncritical routinization of prenatal genetic technologies reinforces this harmful conception of disability, wherein disabled existences ought to be selected against using tools that are increasingly standardized and integrated into expectations of pregnancy management. Taken together, the way societal perspectives of meaningful existence evolve alongside routinized prenatal testing may foreshadow waning protections for disabled communities, a growing neglect for inclusive structures, and a narrower understanding of reproductive justice (Rapp 1998; Roberts 2009; Bridges 2022; Meredith et al. 2023). Framing some lives as intrinsically less valuable than others, without consideration for how societal processes and structures differently position (and disadvantage) individuals, can bear larger concerns around how those with disabilities and their families may struggle to navigate a milieu of institutions regulating social life, resource distribution, and individual accountability for socioeconomic participation and wellbeing.

Technological Imperative and Privatized Health Responsibility in Neoliberal Society

The neoliberal ethos characterizing current socioeconomic systems in the United States has important ramifications on how we interpret pressures to pursue genetic testing, responsibilities around caring for those with disabilities, and healthcare management overall. In the words of political theorist Wendy Brown (2015 p. 9-10, 28), the neoliberal framework "transmogrifies every human domain and endeavor, along with humans themselves, according to a specific image of the economic" that is considered "rational." Though frequently understood within capitalistic political economies, scholars clarify the 'variegated' ways in which neoliberalism– as a "logic of governing" (Ong 2007, p. 3)– presents across social, economic, and political contexts (Brenner et al. 2010). Consistently, neoliberalism bears a focus on individualism, rationality, and prioritization of economic productivity per ableist terms.

The United States' approach to health management—a system wherein individuals are expected to manage their wellbeing via largely privatized access to health insurances and markets, supposedly lowering public health-related economic 'burdens' on the State—is markedly within a neoliberal framework (Rose 2001; Quadagno 2004; Skocpol 1995).[17] Theorizing around this individually-

[17] The United States has a largely privatized healthcare market, yet there are several public healthcare coverage options (although these tend to be far less robust in their coverage). In 2010, the 'Patient Protection and Affordable Care Act' (also referred to as 'Obamacare') was signed into law, representing a significant effort toward providing individuals greater publicly funded access to healthcare and related protections for coverage in the United States. This was the largest expansion of public healthcare coverage since Medicare and Medicaid were passed in 1965 ("Affordable" n.d.). While Obamacare significantly improved access to healthcare coverage, especially in states like California which strongly supported these efforts, there remain issues with robust access to care and political efforts to undercut these measures across many states ("Fact" 2023). As such, healthcare in the United States, even granted public

focused healthcare approach, Clarke et al. (2003) discuss "biomedicalization" as a distinct shift from the prior era of "medicalization." Per medicalization, "aspects of life previously outside the jurisdiction of medicine (came) to be construed as medical problems." However, Clarke et al. (2003) argue that we have now moved onto the era of biomedicalization, which is shaped in large part by our growing reliance on technological developments. This shift toward biomedicalization results from "major, largely technoscientific changes in biomedicine," which fundamentally intertwine technological innovations with our approach to managing health and our embodied identities. We can conceive of the contemporaneous biomedicalization era as medicalization occurring (and shaping our identities and lived experiences) in new and complex "technoscientifically enmeshed ways," compared to medical eras past when technology was less present in our daily lives and less routinized in health management (Clarke et al. 2003, p.161-2).[18]

As biomedicalization plays out within the neoliberal state, its constituent processes shape individual identity and what it means to be a 'responsible' participant in social and economic life. Importantly, notions of 'responsible' or 'valuable' biocitizenship in the neoliberal context— which celebrates technophilic, economically productive individuals—provide a consequential backdrop for the changing understanding of disability and desired existence (Roberts 2009). Ong (2006) describes neoliberalism as having 'mutated' what represents valuable citizenship to idealize "notions of self-efficacy," embodied independence, agency, cognition, and self-control, establishing these as norms for how individuals ought to participate in society— and moreover, the capitalist workforce. Those outside this norm are considered disabled in this narrow iteration of meaningful existence, and "are viewed as lacking the properties needed to be an appropriately functioning and independent individual" able to contribute to economic growth (Sanders and Rogers 2001, p. 489). Here, one's 'appropriate functioning' hinges on an ableist understanding of physical and cognitive independence, with those who are disabled assumed to be perpetually dependent on social support and therefore economically unproductive burdens to the State. This framing of disability focuses on disciplining bodies into normative understandings of embodied ability (to partake in economic productivity), rather than considering how disabilities are produced and exacerbated within social structures and contexts (Russell 2002; Moser 2006; Mitchell 2015). Importantly, this limited understanding of disablement ignores the value of supporting diverse existences and the reality that independence is almost always achieved (even among those not considered typically disabled) through some level of structural and/or technological assistance.

coverage options, remains reliant in large part on individualized measures and resources, placing healthcare funding burdens on individuals more so than the State (Williams and Collins 2016).

[18] Relevant to prenatal genetic technologies being incorporated into pregnancy management, biomedicalization includes processes that routinize technologies into our health practices. These processes consist of the industrialization of medicine, amplified technological reliance (especially for risk surveillance), democratization in the production, distribution, and consumption of biomedical knowledge, and transformations of bodies and identities regarding what it means to be 'healthy' (Clarke et al. 2003, p. 166). Importantly, patients in the biomedicalized era are expected to readily use medical technologies and be informed partners alongside medical experts (Rose 2001). As this chapter will illustrate, processes characterizing biomedicalization take place more sharply in a neoliberal context such as the United States, wherein the responsibility toward health and family are privatized to individuals. Here, though biotechnologies to support health and wellbeing are abundant, public support for disability inclusion are lacking and individuals must largely rely on private resources to manage their healthcare needs (Roberts 2012).

In reshaping valuable biocitizenship, biomedicalization in the neoliberal context also shifts the responsibility for health risk management from the State to individuals, amplifying expectations that patients need to be informed partners alongside their doctors (Rose 2001; Clarke et al. 2003). In prenatal genetic testing, privatized responsibility is represented as pregnant people's social and moral obligation to utilize resources, including testing technologies, to ensure their fetus' health (and help doctors manage their pregnancies accordingly). Physicians increasingly recommend and conduct prenatal genetic testing to obtain insight into obstetric and fetal health, and to increase expecting parents' knowledge and control in antenatal decisions (Bayefsky and Berkman 2022). As patients and providers pursue prenatal genetic testing to assess the health and genetic 'normalcy' of a fetus, these test findings can determine which pregnancies are continued or terminated. Parents' approach to these pregnancy decisions, and the way providers guide parents in doing so, are often (even implicitly) contoured by assumptions about 'responsible' parenthood as it relates to ensuring a child's health and precluding disabilities (Meredith et al. 2023; Piepmeier 2013; Zhang 2020; Asch 2002; Parens and Asch 1999). Moreover, in defining a 'responsible' parent-child relationship (which carries material and social consequences), courtrooms, social services, and healthcare organizations often suggest or strongly imply that parents *should* use their financial resources to access reproductive technologies to ascertain their to-be child's health and genetic status (Solinger 2001; Kritcheli-Katz 2012). The private responsibility to use technologies is further pronounced when the neoliberal state itself offers prenatal genetic testing, as in California's Maternal Serum Alpha-Fetoprotein Screening Program (MSAFP), or through the routinization of testing when considered relatively risk-free, as with non-invasive prenatal testing (NIPT, also referred to as cell-free DNA testing) (Bayefsky and Berkman 2022). Here, the private onus to test pregnancies toward securing health and normative ability for to-be children is pronounced and embedded in the integration of prenatal technologies into standard pregnancy care.

Historically, prenatal genetic testing has been framed as a person's obligation toward society because it is seen as alleviating the State's economic burden of caring for sick and disabled children and adults, consistent with neoliberal principles (Waggoner 2017). Indeed, in the 1970s and 1980s, medical genetics – then framed as an endeavor toward "improvement of the collective gene pool" – was infused into public health agendas (Minna Stern 2012, p. 25). Public health messaging around pregnancy at the time emphasized that parents and providers should use reproductive genetic technologies to prevent births of children with genetic conditions and disabilities. Early justifications for prenatal testing touted a cost-benefit analysis to encourage individuals' supposed responsibility toward the state; they insisted that prenatal genetic testing to identify and terminate fetuses with Down (trisomy 21) and Edward (trisomy 18) syndromes would yield annual savings of up to $66,000, based on precluding the existence of those considered economically unproductive and dependent on the state (Minna Stern 2012, p. 25). Still today, pressures to prenatally test continue (Rapp 1998, Denbow 2015, Waggoner 2019) and disabled individuals in particular are discouraged from family-building, receive substandard prenatal care, and face increased surveillance and stigmatization from states and medical institutions when they do reproduce (Bowen 2022; Piepmeier 2013; Lalvani 2016). Currently, 37 U.S. states allow "disability, as an identity status, (to) be the legal grounding to terminate parental rights," reflecting a priority to reproduce normatively able-bodied citizens (Fritsch 2017, p. 247). Under a privatized health system, where testing is positioned as a 'rational' and almost obligatory part of reproductive decision-making, individuals may choose—or be compelled—to use prenatal genetic testing in

lieu of the State's provision of inclusive structural resources (Lippman 1992; Asch 2002; Parens and Asch 1999).

Along these lines, pregnant people—especially women—face immense social pressure to make 'scientifically sound' decisions on behalf of their fetuses (Rapp1998). Rima Apple (1995 p. 161-2; 2014, p. 116) describes these processes occurring as early as the mid-1800s in the United States, noting how mothers were increasingly expected to be knowledgeable in science, medicine, and technology to raise healthy children. Throughout the 1900s, public health advertisements, widely distributed media, and medical educational materials provided to pregnant women urged them to neglect 'antiquated' generational advice and turn to the 'progressive' scientific guidance of doctors (Apple 1995, p. 164-71). With more medical innovations over time, relying on doctors and embodying the 'Scientific Mamma' involved engaging with technologies aimed at producing children who were healthy and able-bodied according to medical norms. This growing imperative to use prenatal technologies also highlights contentious reproductive politics, where increasingly visible, ever-vulnerable fetuses become the locus of care, rather than those who are pregnant (Ikemoto 1992). In effect, the obligation of ensuring fetal wellbeing (often through prenatal genetic testing) is placed on the pregnant person as the duty of a 'good mother' (Armstrong 1998; Rapp 1998; Denbow 2015, p.109; Waggoner 2019). As this chapter will illustrate, moralizing parenthood in this way is increasingly invoked as prenatal genetic technologies continue to be innovated and integrated into pregnancy care, changing the bounds of upstanding neoliberal biocitizenship as it pertains to health, reproduction, and family-building (Roberts 2012).

Constructing Disability along the 'Promises' of Prenatal Genetic Testing

Disability has been envisioned differently by advocates, disabled communities and their allies, and scholars over time. Undoubtedly, prenatal genetic technologies have had an effect on not only how disability is construed but how disabled communities have faced marginalization as such technologies are often framed as diminishing their very existence (Zhang 2020; Asch 2002; Parens and Asch 1999; Meredith et al. 2023).[19] Below, I discuss various iterations of how disability has been framed, ultimately emphasizing the potential of the 'relational model' of disability as it makes room for how reproductive technological innovations shape what it means to be disabled.

Early conceptions of disability were grounded in understandings of a statistical average–a 'norm'–and deviations from this expectation. Here, disability was seen as an aberration from normalcy or the 'average' person, framed within eugenics ideals of white, middle-class, able-bodiedness (Davis 1997). Today, this approach is reflected in (now sharply contested) medical models of disability, wherein disability is considered an essentialized or embodied individual shortcoming that requires medical interventions to "fix" the body in order to align it with normative expectations of how one

[19] Researchers often use the term 'expressivist objection' to refer to the idea that using reproductive genetic technologies to prevent the birth of disabled children inherently frames disabled life as negative and less valuable. However, recent scholarship suggests that this 'expressivist objection' does not necessarily hold across disabled communities and their families. That is, disabled people and their families have various interpretations of how reproductive genetic technologies should be used toward health and disability outcomes, and many broadly support these tools. While some uphold the expressivist objection, others reject or reframe it depending on their context. Views on the expressivist objection are mediated by the severity of the condition, the broader sociocultural context around disability in a given place, and how the future of disabled people is subsequently imagined – largely representing a 'relational' approach to modeling disability. (Boardman and Thomas 2023; Galis 2011; Winance 2016; Moser 2006)

should exist (Crossley 1999, p. 649). This medical model understands physiology–one's body and genetics–as determinative of their possible life experiences and outcomes. An alternative, the social model, denaturalizes disability from the body, understanding disability as a "social construction shaped by […] physical characteristics built into the environment, cultural attitudes and social behaviors, and the institutionalized rules, procedures, and practices of private entities and public organizations" (Scotch 2000, p. 214). Here, "[disability] is the result of social discrimination, disabling barriers and disablist politics," and interventions necessarily involve social change (Moser 2006, p. 375; Winance 2016). While the social model has been criticized for under-emphasizing the impact of physiological impairments, it is central to many patient advocacy movements, especially among communities mobilizing for greater social recognition and structural support for those living with genetic conditions and disabilities (Navon 2019).

A third, and perhaps more encompassing perspective when analyzing disability alongside technological developments, reframes disability as a 'relational model.' Based on actor-network theory (Latour 2005), this approach to disability traces the *relationships and processes* that co-produce dis/abled subjectivities.[20] Here, one's relative ability emerges out of a social network configuration among individuals, objects (e.g., technologies), and institutions. Importantly, as a part of a variable configuration, one's relative ability is subject to contextual change. In other words, individuals can be considered more or less disabled based on the context in which they are embedded, which can consist of factors such as adaptive institutional structures, resources and communities, and technological innovations (Galis 2011; Winance 2016). Pushing back on ableist benchmarks of embodied autonomy and agency, the relational model fundamentally problematizes the idea that any individual can independently resemble these qualities. It suggests that ability and independence are relative for *all* individuals and conditional on one's embedded social arrangements and larger context (Moser 2006). Criticizing independence and agency as objective concepts, the relational approach to disability also borrows from feminist theories which underscore that "vulnerability is a shared condition," and that individuals are necessarily constrained by "complex social and historical contexts" (Winance 2016, p.105-8).

Per this relational model, physiological disabilities emerge and are exacerbated as a result of shifting structures and relationships. That is, depending on one's surrounding social network configuration, certain characteristics can become more impairing than they otherwise would have been in alternative contexts (with implications for how societally 'valuable' or 'productive' an individual is considered within their context). Along these lines, the value placed on diverse existences changes with sociocultural zeitgeists and institutional practices reinforcing and reflecting notions of which lives are considered meaningful within a given space. These changes are especially visible in the legal realm. For instance, Viviana Zelizer (1981) establishes that markets and health systems change their valuation of children's lives based on contemporary labor structures, as evidenced in the adjudication of children's wrongful death cases over time. Other

[20] Bruno Latour (2005) develops the 'Actor-Network Theory' as an ethnomethodological approach to understanding individual and institutional relationships. The main emphasis of this theory is to analyze associations and connectors between agents and structures, in order to uncover the processes through which institutions are constructed and reified. Relevant to construing technologies as actors in the context of disability, Latour considers objects to be agents with power, agency, and connections. This framework enables the relationships humans share with non-human objects (e.g., technologies) to become 'observable' and more critically interrogated. (Latour 2005, p. 68-69, 72-73, 76).

research reveals how courts recognize and treat disability differently based on shifting societal attitudes around diverse existences (Lejeune and Ringelheim 2019; Mor and Pikkel 2019).

Technology occupies a crucial role in the actor-object network of relational disability, and importantly shapes how disabilities are perceived over time. In prenatal genetic testing, technology stands as both "a mechanism to redress disability" as well as a tool to "force the classification of body as disabled" (Blume et al. 2014). Specifically, prenatal genetic insights transform and "reproduce boundaries between abled and disabled," by designated certain fetuses as 'normal' per their genetic constitution while others are seen as 'abnormal' or 'anomalies' based on their genetic variations (Moser 2006; Navon and Eyal 2016). Healthcare providers–especially reproductive physicians–largely understand the possibility of a fetus being disabled as an undesirable outcome of testing, framing a fetus' medical conditions as essentialized abnormalities or defects. In patient-physician interactions, prenatal genetic technology is often used to highlight the disabled fetus as a "tragic" or "burdensome" event that should be avoided. Parents (especially expecting mothers) who forgo use of prenatal technologies, or carry a fetus with disabilities to term, can also be framed as irrational or irresponsible in their subversion of expert care (Rapp 1998; Piepmeier 2013; Blume et al. 2014; Denbow 2015; Lalvani 2016). Further, obstetricians and gynecologists (ObGyns) often characterize the experience of raising a child with disabilities negatively (Biesecker 1998); a 2016-2021 study revealed that over 60% of ObGyns did not provide disability educational materials to patients following a fetal Down Syndrome diagnosis (Meredith et al. 2023).[21] Consistently, patients receive negative—rather than supportive—information regarding raising disabled children following prenatal testing, when indeed research has shown that with adaptive supports and suitable structures raising a child with disabilities is no less fulfilling (Minear et al. 2015). Given the social importance accorded to physicians, their guidance on using prenatal technologies is often reinforced as the 'correct' or 'rational' choice. Consequently, many pregnant people rely on physicians' views (often reflecting the medical model of disability) during testing and reproductive decision-making, illuminating the significance of physicians' interpretation of technologies and genetic insights on parent's views on disabilities and their ability to care for a future child (Bernhardt et al. 1998). (Asch 2002; Parens and Asch 1999)

The conceptualization of disability as evinced by actor-object networks, systems, and interactions surrounding prenatal genetic testing technologies reflects biological determinism and moralized expectations of parenthood. Within a relational configuration that includes prenatal testing to classify pregnancies as normal or abnormal, and medicalized interactions that reify these harmful notions, disability is discussed as pertaining to the body and parents as those responsible for managing such consequences prenatally. Here, one's quality of life is seen as genetically rather than socially rooted, ignoring the significant degree to which social factors determine one's quality of life. When technology serves to categorize some existences as abnormal and unwanted based on genetic differences as a source of disablement, this absolves the State's responsibility toward fostering inclusive environments and neglects the powerful influence of developing equitable social structures (Mukherjee and Shirinian 2022; Comfort 2018).

[21] These statistics are consistent with those from past decades. A 2008 study similarly found that over 70% of ObGyns did not provide disability education to patients following a fetal Down Syndrome diagnosis (Minear et al. 2015).

The reluctance of neoliberal states and their medical institutions to frame disability as a relational construct reinforces the "logic that individuals are primarily responsible for their own fates" and that "families and voluntary agencies, rather than local states, should bear the onus of responsibility for assisting persons in need" (Fritsch 2017, p. 254-5, 261). When disabilities are seen as an embodied personal shortcoming, individuals bear the private cost of disciplining their bodies into those considered autonomous and valued by an economically driven state, rather than states meaningfully adapting the contexts in which individuals are embedded. Through examining the neoliberal state (via its legal institutions) and routinized prenatal genetic technologies as a part of the assemblage of actors, objects, and institutions comprising disability, this chapter situates disability as a mutable and relative identity. It emphasizes how socioeconomic and political configurations contour the interpretation of biotechnologies, informing the extent to which those with disabilities are understood and differently valued as biocitizens.

The Role of Tort Law in Transforming Disabilities within Neoliberal Society

Contemporary economic structures, influential social organizations, and dominant sociocultural values situate how courts adjudicate cases. Courts both reflect these contexts while dialectically shaping them, which can be observed through their judgements and arguments offered in support of their legal conclusions. With regard to cases pertaining to disabilities, many laws in the United States continue to uphold a medicalized conception of disability, one that positions disability as an embodied shortcoming in need of medical treatment, despite the existence of the Americans with Disabilities Act (ADA). Signed into law in 1990, the ADA is perhaps the most significant legislation protecting people with disability against discrimination. It signals a policy commitment to the social inclusion of people with disabilities and enshrines an understanding that societal structures can and should be inclusive of those with varying needs (Scotch 2000, p. 216). However, despite the ADA, the existence of wrongful birth and life torts and the Health Security Act continue to emphasize the prevention of diseases, those with disabilities, and inherited conditions rather than inclusive structures (Russell 2002). Under these legal frameworks, prenatal genetic testing is a tool to preclude the birth of disabled babies, where disabilities are seen as embodied legal harms caused to individuals and families (Morris 1994, p. 304). Within a historical and socioeconomic context promoting economic productivity, independence, and privatized responsibility, the way WBL torts are adjudicated in the United States largely echoes a medical understanding of disability during prenatal testing.[22]

The elements of WBL tortious claims are similar to typical medical negligence cases; they involve: (i) a duty of medical care, (ii) a breach of this duty, and (iii) a causal relationship between violation of this care standard and the harm sustained. The healthcare provider may not have directly caused the harm sustained by the child (i.e., their disability), but their (in)actions are the *proximate* cause of this outcome. Wrongful birth and life claims differ mainly in terms of who brings the claim (i.e., plaintiff) against the medical provider (i.e., defendant), and how the harm sustained is framed. Wrongful birth claims are brought by the plaintiff-parents against the healthcare provider for the

[22] Tort law provides a fitting venue for exploring how neoliberalism contours biomedicalized reproduction and the ongoing conceptualization of disability and valuable existence. In torts, responsibility for injuries must be ascribed to specific individuals. If an injury's cause cannot be attributed to an individual, it is nothing more than misfortune, and compensation is not available. In this way, tort law obscures unsuitable social structures and their role in exacerbating inequalities, leaving them unchallenged as it places the onus on individuals (Mor 2014). A tortious understanding of wrongdoing and accountability with regards to disabilities and prenatal genetic testing resonates with the especially individualistic ethos of neoliberalism. (Dorfman 2017; TenBroek 1966; Hensel 2005; Mor and Pikkel 2019).

deprivation of their reproductive choice to terminate a pregnancy where a fetus was indicated to have disabilities or genetic conditions. This is fundamental to a WB claim's success: parents must assert that, *but* for the physician's failure to identify and inform them of the fetal condition in a timely manner, they would have terminated the pregnancy. On the other hand, WL claims are brought by the plaintiff-child (through their parents) against the healthcare provider. Here, the negligence is that the parents' deprivation of "the decision to abort or never conceive" led to the child's 'wrongful' life (Beasley 1992, p. 234). The plaintiff-child must establish that it would have been preferable for them to have never existed rather than to exist in their current disabled state, which has larger societal consequences for how we consider lives worth bringing into existence.

Along these lines, a main "stumbling block" for courts contending with WL claims is recognizing the child's disability as a legal injury, which bears implications for how various lives and existences are (de)valued as representing 'harms.' Without recognizing the child's disabled existence as an injury, however, courts are not able to calculate or award damages to the plaintiffs (Burns 2003, p. 811). These challenges emerge because tort damages are specifically intended to restore the plaintiffs to their pre-tort condition; in WL cases, the pre-tort condition would be non-existence. However, courts and legal scholars have long argued that comparing disabled existence to non-existence (a counterfactual) is not tenable (Amaral-Garcia 2018, p. 387, 391): "a nonexistent person does not experience the absence of any benefit of creation, and has no life that could be worse" (Shiffrin 1999, p. 134). As such, WL claims can be particularly difficult to adjudicate in terms of establishing provider culpability and damages to infants and their families.

By understanding negligence and legal injuries as the failure to terminate disabled fetuses, WBL torts undermine reproductive justice. Reproductive justice is a "law-focused" and "social justice-aimed" movement that emphasizes an intersectional approach to ameliorating structural inequalities (Luna and Luker 2013). Reproductive justice underscores that the narrow legal approach of 'reproductive rights' is not always a suitable or all-encompassing mechanism to ensuring equity when it comes to reproduction and valuing diverse existences. It problematizes a rights-based conception, wherein one's bodily autonomy is typically celebrated only in terms of being able to terminate unwanted pregnancies. Instead, the reproductive justice framework seeks to uphold reproductive choice as a whole, including the choice to bring to term and parent children with disabilities and various conditions. It also expands the idea of parenting 'rights' to include "the right to have children" for all individuals (e.g., including low-income or otherwise marginalized communities) and "the right to parent with dignity" (e.g., for incarcerated parents, parents with disabilities) (Luna and Luker 2013; Bailey and Peoples 2017; Roberts 2009). WBL torts undermine reproductive justice because they emphasize the *right* to terminate *specifically* fetuses indicating disabilities, suggesting that carrying such pregnancies to term constitutes a larger societal harm. WBL claims reinforce parents' choice to birth a particular ('normal' or 'productive') child; that is, to avoid parenting children with disabilities. Not only does this prioritize some existences as more valuable than others, but it also assumes all pregnant people have access to the same reproductive opportunities, disregarding how social structures mediate one's socioeconomic positionality and available reproductive options (Patton 2000; Piepmeier 2013).

Tort law is also discussed as "the law of disablement" in how it shapes (and stigmatizes) plaintiffs' identities into becoming disabled (Mor and Pikkel 2019, p. 30; Bloom and Miller 2011, p. 714). Tortious claims are constructed through a process of "naming, blaming, and claiming" (Felstiner,

Abel, and Sarat 1981, p. 181). In WBL cases, 'naming' an injury depends on how disability is defined. As plaintiffs' bodies are placed on "on trial," they must be identified as "incomplete" in order to be understood as disabled (Bloom and Miller 2011). In this way, naming disability as a harm has been described as a "[spectacle] of misery," where plaintiffs must be "willing to openly disavow their self-worth and dignity" to establish their disabled existence as a less meaningful life (Mor 2014, p. 31; Hensel 2005, p. 171). Plaintiffs are incentivized to rely on negative cultural stereotypes and the medical model of disability, nullifying the value of disabled life and framing it as an unwanted injury (Sheth 2006, p. 648). After disability has been named as a harm, liability for these injuries is then ascribed to individuals who are 'blamed,' privatizing the responsibility for supporting those with disabilities. Finally, through their 'claims,' plaintiffs are awarded compensation for their injuries (i.e., disabilities). Through claiming (i.e., assessing damages to be awarded), the disabled body is often compared to the 'healthy' 'intact' body, further reinforcing the inequitable medical model where disabilities are embodied and essentialized (Felstiner, Abel, and Sarat 1981). While courts alone cannot transform broader understandings of disability, through these processes they are fundamental in defining the societally appropriate use of biotechnologies and how these tools shape the classification of disabilities as undesirable existences.

This chapter bridges the neoliberal state's socioeconomic values, technological co-construction of disabilities, and dialectical relationship between law and society. In doing so, it argues that– when interpreted within a neoliberal context– developments in prenatal genetic technologies can cast disabilities as unwanted existences and amplify caring for those with diverse needs as private burdens. Analyzing WBL cases, it shows that the biomedicalization of reproduction– i.e., routine use of prenatal genetic technologies during pregnancy– shapes values around reproducing those with disabilities. Overall, the chapter illuminates how legal institutions co-construct an understanding of disability that relies on narrow notions of bodily independence and socioeconomic productivity, underscoring the use of prenatal genetic technologies to prevent disabled lives, privatizing family health responsibilities, and considering certain existences inherently less meaningful.

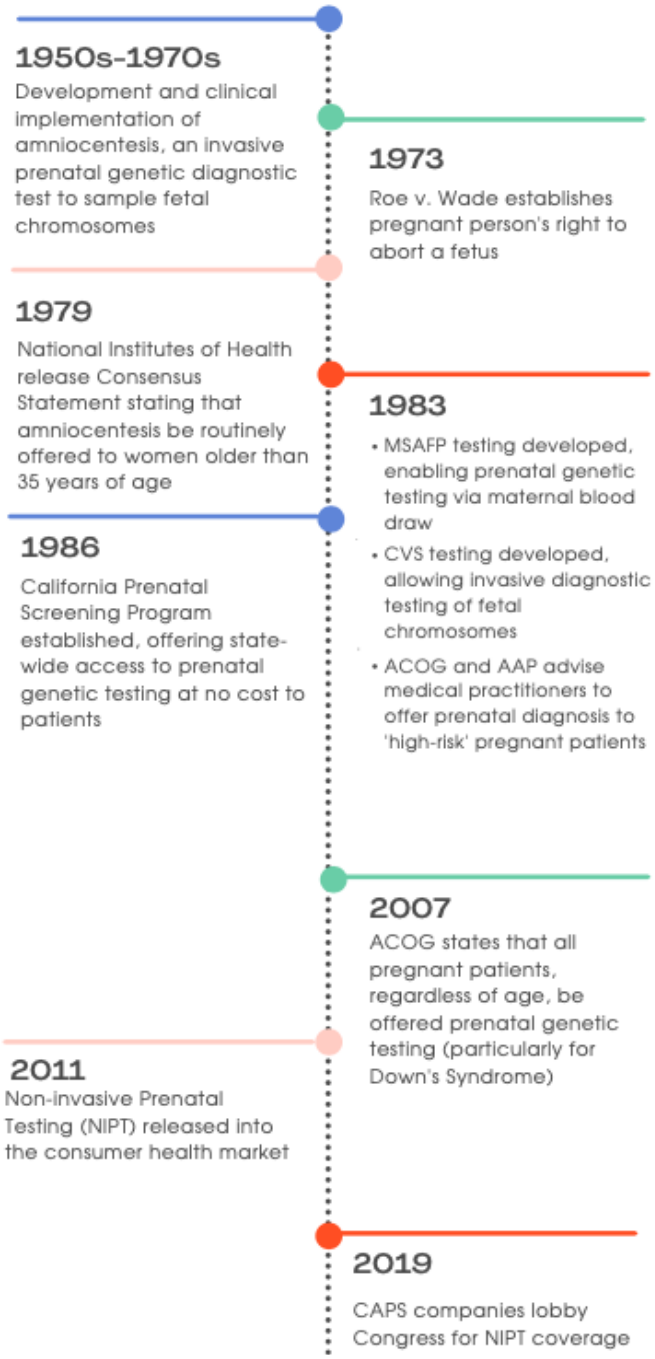
Overview of Prenatal Genetic Testing Technologies and Relevant Policies

Prenatal genetic testing technologies have evolved substantially over the last half century. Their development has shaped reproductive practices and decision-making in the United States, with several public initiatives helping routinize their use during pregnancy management. In 1982, the American College of Obstetricians and Gynecologists (ACOG) and the American Academy of Pediatrics (AAP) advised physicians to offer prenatal diagnosis (e.g., amniocentesis, chorionic villus sampling) or provide a referral to such services for medically appropriate candidates (e.g., pregnancies at high-risk for genetic conditions). Shortly thereafter, in 1986, California launched the California MSAFP Screening Program, offering prenatal screening services and pledging state-wide access to prenatal genetic testing. In 2007, ACOG stated that *all* women—regardless of age or other reproductive risk factors—should be offered prenatal genetic testing for fetal genetic aneuploidies (ACOG Practice Bulletin 77 2007, p. 217; ACOG Practice Bulletin 88 2007, p. 1459). More recently, in 2019, the Coalition for Access to Prenatal Screening (CAPS), an alliance of four genetic technology companies, lobbied Congress to provide Medicare and Medicaid funds to cover access to NIPT, situating it as a key component of public healthcare.[23] Today, with both ACOG

[23] CAPS consists of four leading genomics companies: Illumina, Myriad Women's Health, Labcorp, and Natera. There stated mission is to "improve access to state-of-the-art prenatal screening using cell-free DNA (cfDNA)-based

and the American College of Medical Genetics (ACMG) recommending providers offer *all* pregnant people prenatal genetic testing via NIPT, the technology has become a standardized part of pregnancy care (Dungan et al. 2023; "American" 2021). Below, figure 1, illustrates these notable developments.

Figure 1: Summary of Key Events in Prenatal Genetic Testing Development



noninvasive prenatal screening (NIPS) […]." They describe a commitment "to encourage appropriate legislative measures and reimbursement coverage policy changes for this medically actionable testing service, which has the potential to improve personalized patient care." ("Our Mission" 2023)

Since the introduction of non-invasive prenatal testing (NIPT) in late 2011, prenatal genetic testing has become more expansive, accessible, and medically easier to conduct. With the increasing implementation of prenatal genetic testing, these technologies bear significantly on reproductive choices, especially with regard to which fetuses are carried to term based on anticipated health and disability status. For example, as discussed, provider-patient interactions around prenatal genetic results that indicate fetal genetic conditions or disabilities often result in implicit pressures to terminate these pregnancies (Denbow 2015; Meredith et al. 2023). WBL disputes provide an avenue where we can observe how emerging prenatal genetic technologies have shaped broader notions of disability, as courts adjudicate the technologies' 'proper' societal use particularly with regard to preventing certain existences. These cases surrounding the use, negligence, and/or miscommunication about prenatal genetic testing represent key instances wherein one's health status is questioned as a matter of desirable existence.

This chapter focuses largely on how prenatal genetic innovations have taken hold in California, including the state's relevant policies and how WBL disputes are adjudicated within this setting. California's legal influence around prenatal genetic testing is salient when it comes to regulations, laws, and practices nationally and internationally. In particular, WBL claims are especially relevant within the Californian setting: California stands at the cutting edge of biotechnology and genetics, simultaneously contending with a fraught history surrounding eugenics practices (Manian 2020; Watanabe 2020). It is home to leading medical institutes where new genetic technologies and innovative medical practices have emerged. California is also a "hub" for assisted reproductive services including prenatal genetic testing (Dunn 2019). Especially relevant to prenatal genetic technologies in the context of WBL disputes, California does not prohibit abortions based on fetal disability, unlike 14 other states that have passed such laws. California is one of few states that has remained committed to maintaining access to abortion. For states with disability-specific abortion bans in effect, including Missouri, North Dakota, and Ohio, prenatal genetic testing cannot influence reproductive decision-making during pregnancy, which is a main consideration in WBL cases (Bridges 2022). As such, the California courts contend with innovative practices, technologies, and social issues not as readily realized in other places.

Transforming the Conceptualization of Disability Alongside Prenatal Genetic Testing

The following empirical discussion provides an in-depth qualitative analysis of WBL cases specifically related to prenatal genetic testing. Using the online database "Casetext," I obtained a sample of cases relevant to California or federal appellate courts; federal judgements were included as these are persuasive as precedents in both state and national cases.[24] This sample included all WBL cases heard at the various court levels in California, including the Supreme Court, Court of Appeals, and District Courts. Judgements that were published between 1963 and 2021 were included in the sample, encompassing changes over the past 58 years; however, a close analysis was undertaken for those cases between 1963 and 2015, as these represented notable developments and precedents in how the use of prenatal genetic technologies has been interpreted alongside notions of disability. The analysis also encompasses landmark and especially influential WBL cases from other states within the United States. In total, 37 cases were analyzed, 16 of which were

[24] Casetext is a comprehensive legal database, freely accessible online, which includes all federal and state cases and statutes above trial court level.

Californian cases. Of the California judgements, 9 were WL claims, 2 were WB claims, and the remaining 5 were WBL claims. Table 1 provides a partial summary of the cases analyzed.[25]

Table 1: Partial Summary of Wrongful Birth and Life Cases Closely Analyzed

Name	Year	Court	State	Facts	Claim	Judgement
Zepeda v. Zepeda	1963	Court of Appeal	Ill.	Plaintiff sued his father for having conceived him out of wedlock and claimed damages for the deprivation of his right to be a legitimate child.	WL	WL claim dismissed in trial and Supreme Court for legal insufficiency of the complaint. Plaintiff cannot sue for their pain and suffering. Court concerned with implications of creating this new tort.
Gleitman v. Cosgrove	1967	Supreme Court	N.J.	Plaintiff-mother alleged doctors were negligent in advising her that her rubella wouldn't harm the fetus.	WB	WB claim denied; courts held nothing could have brought about a perfectly healthy child.
Park v. Nissen	1975	Superior Court	Cal.	Plaintiff-mother was not informed of availability of amniocentesis.	WL	WL denied. Jury found that amniocentesis was not required by community standard of care at the time.
Berman v. Allan	1979	Supreme Court	N.J.	Plaintiff-mother was pregnant at age 38; doctor failed to offer an amniocentesis to check if plaintiff-infant had Down Syndrome.	WB & WL	WL claim denied. WB granted. Court also reversed Gleitman decision, noting *Roe v. Wade (1973)* enables parental choice of abortion.

[25] Note, this table is not an exhaustive representation of all the cases analyzed in this chapter. Rather, it is a 'partial summary' of the cases analyzed in order to illustrate the various issues pertaining to WBL cases undertaken in United States courts since 1963 and provide a sense for the changing outcomes.

Curlender v. Bio-Science Laboratories	1980	Appellate Court	Cal.	Defendants incorrectly informed the plaintiff-parents they were not carriers of Tay-Sachs disease.	WL	WL claim granted: "a plaintiff both exists and suffers, due to the negligence of others." Reliance on *Roe.*
Turpin v. Sortini	1982	Supreme Court	Cal.	Plaintiff-parents claimed defendants inaccurately evaluated their first child for deafness, leading to plaintiff-infant also being born deaf.	WL	WL claim granted. Special damages awarded: "Impaired life is not always more valuable than nonexistence"
Harbeson v. Parke-Davis, Inc.	1983	Supreme Court	Wa.	Plaintiff-mother prescribed Dilantin to control her seizures; specifically asked doctors about possible birth defects and was assured there were none; plaintiff-infants were both born with fetal hydantoin syndrome.	WB & WL	WL claim granted. Special damages awarded.
Andalon v. Superior Court	1984	Court of Appeal	Cal.	Plaintiff-infant alleged doctor's failure to offer amniocentesis caused him to be born with Down Syndrome and forgo earning capacity.	WB	WB denied. Plaintiff-infant did not suffer a legally cognizable injury: "There is no loss of earning capacity caused by the doctor in negligently permitting the child to be born with a genetic defect that precludes earning a living. One

						cannot lose what one never had."
Simmons v. West Covina Medical Clinic	1989	Court of Appeal	Cal.	Plaintiff-mother had a blood test which indicated a 20% risk of Down Syndrome in the fetus, but the doctor did not tell the patient this result or recommend further testing.	WB and WL	Claims denied. Doctor was not negligent; 20% falls far short of the contemporary requisite reasonable medical probability standard of causation.
Reed v. Campagnolo	1993	Court of Appeal	Md.	Plaintiff-parents allege negligent failure to inform about AFP testing, amniocentesis, and abortion.	WB and WL	WL abandoned; WB recognized where doctor does not inform about available possible diagnostic testing.
Gami v. Mullikin Medical Center	1993	Court of Appeal	Cal.	Plaintiff-parents allege damages for negligence, as plaintiff-mother submitted a sample for AFP testing, but no one told her it was unsuitable thus depriving of the opportunity to learn about her fetus' anomaly.	WL	WL and special damages granted.
Galvez v. Frields	2001	Superior Court	Cal.	Plaintiff-parents allege the doctor failed to make reasonable efforts to ensure plaintiff-mother was given a blood test to detect fetal spina bifida.	WL	WL granted, as AFP test for pre-natal screening of neural tube defects seen as care standard: "impaired child may recover special damages for the extraordinary expenses necessary to treat the hereditary ailment from

						which he or she suffers."
Johnson v. Superior Court	2002	Court of Appeal	Cal.	Cryobank failed to inspect whether donor had hereditary kidney diseases prior to approving him, leading plaintiff-infant being born with ADPK.	WL	WL denied. Court recognized that gene in the sperm, and not the defendants, had caused the ADPK. Plaintiff "had obtained a physical existence with the capacity both to receive and give love…as well as to experience pain and suffering"
Barragan v. Lopez	2007	Court of Appeal	Cal.	Plaintiff-infants born with cerebral palsy; plaintiff-mother claims doctor should have advised her of her right to have an abortion.	WL	WL denied. Doctor did not owe plaintiff-mother a duty to advise of right to an abortion because there was no prenatal indication of the condition.
Ermoian v. Desert Hospital	2007	Court of Appeal	Cal.	Plaintiff-infant born with brain abnormalities; claims doctors deprived mother the choice to have an abortion.	WL	WL denied. In 1994 (time of pregnancy), California prohibited third-trimester abortion unless necessary to protect mother.
Wuth ex rel. Kessler v. Lab. Corp. of America	2015	Court of Appeal	Wa.	No genetic counselor for CVS resulted in undiagnosed genetic condition. Plaintiffs falsely reassured of fetal health.	WB and WL	Claims granted, as care standard of counseling neglected. Plaintiffs granted $50 million in damages.

The case analysis ultimately generated three key themes that structure the following empirical sections: changing expectations of prenatal genetic technologies, shifting emphasis on parental responsibility, and the value of disabled existence.[26] Overall, the chapter illustrates how courts, in their adjudication and awarding of damages, position disability as unwanted and consider prenatal genetic technologies to be a solution parents and providers ought to use to preclude such birth outcomes. Further, as the availability of increasingly precise prenatal genetic information enables more fetal conditions to be classified as 'deviations' from normative health and ability, courts too enlarge their parameters around which disabilities and genetic variations constitute legally cognizable injuries. In other words, as prenatal genetic technologies offer augmented insight into more genetic differences, courts come to consider more of these (even mild) variations as disabilities compromising one's life and representing societal harms. This is evinced in how courts interpret technological advancements, concurrent recommendations from professional medical societies, and legislation and policies concerning reproductive choices and fetal existence. Taken together, in their adjudication of WBL cases pertaining to prenatal genetic technologies, courts reason that technological advances *should* be used toward reproducing normative health and ability (a continually shrinking standard), thereby illuminating broader sociocultural shifts regarding how genetic technologies inform reproductive decisions and conceptualizations of disability.

First, this chapter examines the ongoing biomedicalization of pregnancy, showing how 'healthiness' is defined based on what insights contemporary genetic tools can offer. Specifically, it delves into the steady advancements in prenatal genetic testing and the professional guidelines accompanying their implementation. It reveals how testing becomes routinely incorporated into reproductive medicine and gradually reduces the tolerance of 'errors' leading to the birth of babies with disabilities and genetic conditions that *should* have been identified (and precluded) in-utero.

Next, I discuss shifts in parental responsibility around testing and raising disabled children. This discussion also emphasizes the role of *Roe v. Wade* (410 U.S. 113 [1973]), which took effect in 1973 and enabled access to pregnancy termination nationwide. While *Roe* represented a critical step forward in protecting reproductive autonomy (much of which has been reversed given the 2022 *Dobbs v. Jackson Women's Health Organization* decision, revoking the federal right to abortion; see Horn 2022), in the context of WBL cases and prenatal genetic technologies *Roe* is often harnessed to emphasize parents' imperative to terminate specifically pregnancies with chromosomal variations and disabilities.[27] As such, in this context, *Roe* also marks a milestone where pressures to take individual measures (i.e., abortion) to preclude disabled existences were

[26] This was a largely deductive analysis of WBL cases, paying particular attention to courts' rationale and dicta related to awarding damages or lack thereof. The themes used for qualitative coding came from literature about health construction, disability and reproductive justice, gender expectations, and social inequalities related to genomics innovations. As cases were read multiple times, other emerging relevant codes and themes were included. Although the initial analytical approach was deductive, the overall analysis also accommodated inductive findings to best represent the data.

[27] As described, and further explored within the empirical sections below, this development also has notable consequences for compromising reproductive justice, wherein the 'right to terminate' is celebrated and emphasized only with regard to aborting fetuses with genetic conditions or disabilities. In this limited framework of reproductive autonomy, there is unequal and diminished upliftment of the choice to birth disabled children and extend the right to dignified parenthood to individuals of varying (especially marginalized) backgrounds. (Luna and Luker 2013)

amplified. Here, I also illustrate how the neoliberal context contours biomedicalization of pregnancy, showing how children with disabilities are described as 'societally undesirable' and how their care is framed as a private burden for their families.

Finally, I turn to how disability is differently managed and conceptualized over time alongside developments in prenatal genetic testing. The discussion focuses on compensation models that recognize disabled existence due to negligent prenatal genetic testing as a legal injury. I show how these legal dicta and judgements awarding damages gradually position disabled existence as inherently less valuable and objectively undesirable. Throughout, the discussion illuminates how the routine and uncritical use of prenatal genetic technologies – particularly when situated within neoliberal discourses and narrow frameworks of what constitutes meaningful existence – foreshadows harmful implications for reproductive protections, disability justice and inclusion, and frames some lives as 'abnormal' and thus intrinsically unwanted.

How Technological Promises Transform Expectations in Reproduction Management

Over the decades, WBL cases illustrate how biomedicalization– specifically, routinization of prenatal testing in pregnancy care– unfolds and the implications this has on medicalized reproduction and disability discourses. In the cases discussed below, we see how technologies are developed and made more precise over time, gradually becoming a standard part of pregnancy management. There emerges an idea that, as prenatal genetic technologies become more available, we should be able to increasingly rely on these tools to enable certain (ableist) visions of meaningful existence. Courts start to see these technologies as tools that parents, and societies more broadly, should be able to turn to in order to prevent birth outcomes of children with disabilities or genetic conditions. The development and acceptance (even encouragement) of prenatal technologies as routine pregnancy management changes expectations around how these tools *should* be used and the types of health and life outcomes we *should* aim for. Over time, as courts adjudicate matters of technological innovation, values around reproductive decisions and family building are encoded into policies and legal precedents, which come to represent larger societal priorities around various existences and the 'right' way to approach reproduction based on routinized genetic technologies.

Prior to prenatal genetic insights, there was little liability on ObGyns for failing to inform expecting parents about their genetic carrier status or potential fetal conditions, as these were challenging to determine prenatally without genetic tools. However, advances in genomic research and technological innovation increased the breadth and precision of genetic data about fetal health during reproduction (Bayefsky and Berkman 2022). As this expanded technological capacity boasts more sensitive, comprehensive, and reliable genetic test results, and health experts begin recommending testing accordingly, court judges reflect the sentiment that these tests should be an expected (yet optional) part of pregnancy care. Judges further emphasize that individuals should be able to anticipate increasingly accurate results from these prenatal genetic technologies to make reproductive decisions (e.g., pregnancy termination). Eventually, courtrooms also expand physicians' duties to encompass prenatal genetic testing. They move from tolerating more risk, when testing technologies were nascent, to narrowing the room for error around identifying a fetus' potential genetic condition or disability as these genetic technologies are further developed and integrated into pregnancy care. As genetic technologies and related health surveillance are steadily standardized into typical pregnancy care, courts shape collective ideas around heightened

expectations to use these technologies and largely consider these innovations to be societally valuable in their potential to preclude genetic conditions and disabilities.

Before professional societies and medical organizations released statements recommending use of prenatal genetic in the mid-1970s, such testing was not widely accepted as a part of routine pregnancy care. As such, there were fewer expectations placed on reproductive physicians to provide genomics-related medical care to pregnant patients. In the early case of *Gleitman v. Cosgrove*, 227 A.2d 689 (1967), the judiciary reflected this approach. Here, the plaintiff-parents sought damages from the defendant-physician, as their son was born with severe congenital challenges resulting from his mother contracting rubella in the first trimester of her pregnancy. However, since prenatal genetic testing was not widespread at the time, and abortion was not possible (leaving no suitable post-conception action), the courts refused to recognize the claim or compensate for "the intangible, unmeasurable, and complex human benefits of motherhood and fatherhood." Similarly, in *Park v. Nissen*, No. 190033, Cal. Super. Ct., Orange Cty., Dec. 13, (1974), the jury decided that amniocentesis—and prenatal diagnostic testing more widely—was not considered "a required community standard of care" at that time. Therefore, in in *Park v. Nissen* (1974), the defendant-physicians could not be faulted for the child in this case being born with Down Syndrome; there was no contemporary expectation that physicians conduct prenatal genetic testing in order to ascertain a fetus' chromosomal variations.

As *Gleitman* (1967) and *Park* (1976) show, in the 1960s and part of the 1970s, not only was prenatal testing outside standard care, but reproductive physicians were not yet expected to engage with genomics in this way. We also see this reflected in *Howard v. Lecher*, 53 A.D.2d 420 (1976), where the plaintiff-parents claimed that the defendant-physician was negligent because he had not taken a proper medical history despite the plaintiffs being of Eastern European descent (and thus potential carriers of Tay Sachs). However, the court did not consider physicians' "scope of duty" to include being "forced genetic counselors,"[28] and the plaintiffs' WB claim was unsuccessful. As such, at the time, ObGyns' responsibilities did not encompass providing patients prenatal care aligned with the ongoing advances in genomic knowledge and technologies.

However, as amniocentesis became accepted and standardized throughout the 1970s, courts were more willing to recognize medical malpractice where pregnant parents had not been accurately or adequately informed about prenatal genetic testing and counselled about related risks regarding their fetus' genetic health. This marks a notable departure from earlier WBL cases, which were often dismissed due to an "inability to find that the infant is worse off as a result of the negligence […] than had the infant never been born" or "that the fetus could not be legally aborted at the time." Pushing back on the precedent of such dismissals, *Park v. Chessin*, 60 A.D.2d 80 (1977) emphasizes that "cases are not decided in a vacuum; rather, decisional law must keep pace with expanding technological, economic and social change." In *Park v. Chessin* (1977), the plaintiff-infant was born with infantile polycystic kidney disease and sought damages from the defendant-physicians. She claimed physicians had misinformed her parents that her condition was not hereditary, thereby precluding them from making an informed decision about her birth. In 'keeping pace' with societal developments, the *Park* intermediate level appellate court found the plaintiff's claim to be judicially cognizable; the defendant-physicians' negligence around appropriately

[28] *cf.* Naccash v. Burger, 290 S.E.2d 825, 837 (Va. 1982).

counseling pregnant parents about their reproductive genetic risks, and not using genetic tools toward this end, constituted a "breach" that foreseeably resulted in that child's wrongful birth.

Park underscores that changing federal abortion laws in the early 1970s not only afforded "potential parents the right […] not to have a child," but that this right "extends to instances in which it can be determined with reasonable medical certainty that the child would be born deformed" (60 A.D.2d 80 at 88).[29] In other words, parents should be able to specifically prevent the birth of children with disabilities or genetic conditions using prenatal genetic technologies, reinforcing the idea that there is a 'correct' way to use these tools toward societal reproduction. Further, though the *Park* court did not specifically define 'reasonable' certainty, it laid down the expectation that a child has a "*fundamental right* […] to be born as a *whole, functional* human being," with disabilities, in contrast, representing incomplete and dysfunctional existence when compared to this model of embodied ability and health (*emphasis added*). The judgement emphasized the idea that medical negligence in prenatal genetic testing and genomic information can deprive one of this so-called birth right. Accordingly, by the late 1970s, courts explicitly interpreted reproductive autonomy legislation and disabilities alongside the imperative to rely on technological advancements. In doing so, they began to shift and reflect the grounds for what parents (and wider society) should expect from prenatal technologies with regards to their child's normative health and ability.

Soon after *Park v. Chessin* (1977), prenatal testing usage increasingly became an expected part of care, as recognized by the 'Consensus Development Statement' released by the National Institutes of Health (NIH) in 1979. This statement recommended that providers offer amniocentesis to any pregnant person over 35 years of age or with any other risk factor. In addition to amniocentesis (which can diagnose fetal genetic conditions), screening via MSAFP (testing for maternal serum alpha-fetoprotein, which provides risk-based likelihoods that a fetus has certain genetic conditions) soon followed as routine care expectation. While ACOG initially stated that "routine MSAFP screening of all [pregnant women] is of uncertain value," based on concerns about insufficient availability of related services including high-quality laboratory processing, genetic and reproductive counselling, follow-up ultrasounds, and confirmatory amniocenteses, its Department of Professional Liability soon reversed this position (ACOG 2007a). Following multiple obstetric malpractice suits, ACOG issued the 'Professional Liability Implications of AFP Tests,' which recognized this test as falling well within the standard of care in prenatal clinical management. Shortly thereafter, the MSAFP screening was cemented by the 1986 California Prenatal Screening Program, wherein the state pledged to cover access to prenatal screening, and related genetic tests, for *all* pregnant people. Still in effect today, this program requires that MSAFP screening be offered to *every* pregnant patient, who then signs statements of informed consent or refusal, thereby maintaining voluntary participation even while routinizing prenatal genetic testing as public health infrastructure (Steinbrook 2002). That same year, the 'California Health and Safety Code' (§289.8) also required clinicians to advise all pregnant patients of this blood test for AFP that can indicate a fetus' potential genetic conditions. In this way, we see how expectations around technological developments go hand in hand with public health measures and guidelines for medical

[29] *Roe v. Wade* was signed into law in 1973 and provided legal access to abortion across the United States. This decision was effectively undone in 2022, in the *Dobbs v. Jackson* case. The impact of abortion laws is discussed further in later discussions within this chapter.

professionals, working in tandem to routinize prenatal genetic testing in one's medicalized pregnancy care experience.

By the early 1990s, prenatal genetic testing was well on its way to being routinely used in California clinical settings. At this point, Physicians were typically encouraging patients to pursue the state-run MSAFP screening and implementing it as standard practice, rather than informing them about its purpose and voluntary participation (Rothenburg and Thomson 1994, p. 206). This routinization of prenatal genetic technology presupposed that pregnant people were able to make free and informed choices around their reproductive decisions. However, the choice between screening and not screening, and—more importantly, between termination and continuation of a pregnancy possibly affected by genetic conditions—was not necessarily available to all individuals. As genomics was relatively new to public knowledge, many individuals had limited understanding about the process of prenatal genetic testing and the implications of its results, and genetic counseling was not yet a widespread medical provision to fill such gaps in patient education. Several scholars have argued that, at the time, routinized prenatal testing was less about providing pregnant individuals with informed choices, and more to do with the systematic de-selection of fetuses with 'unbearable' conditions (Rapp 1998; Garland-Thompson 2015).

WBL judgements change as prenatal testing expectations become routinized and genomic medicine is integrated into physicians' professional scope of duties. Take, for example, *Becker v. Schwartz*, 46 N.Y.2d 401 (1978), where the plaintiff-mother, a 37-year-old woman, was never advised of the availability of amniocentesis to test whether her fetus had Down Syndrome. She was successful in the WB suit against her physician for his failure to refer her for an amniocentesis; she was able to establish that, because she met the criteria of 'advanced maternal age' (above 35-years-old), she fell into the category of pregnancy people who should be offered testing per concurrent medical recommendations. Importantly, the *Becker* judgement defined physicians' liability as failing to inform patients of their reproductive risks and the tests and procedures available to mitigate such risks. Unlike earlier cases, physicians' affirmative scope of duties and the standard of care were now concretely expanded to encompass investigating genetic factors and informing parents of these results and related risk reduction measures.

With the development and routinization of prenatal genetic technologies, courtrooms also emerge as important sites for defining acceptable thresholds for risk tolerance and medical error. Tolerance for risk and error is especially pertinent to prenatal genetic testing, which provides risk-based results and can engender uncertainty regarding the child's eventual health outcomes (Kliff and Bhatia 2022; discussed further in the following chapters). In a relatively early case, *Simmons v. West Covina*, 212 Cal. App. 3d. 696 (1989), the defendant-physicians administered AFP testing to the plaintiff, indicating her fetus was at 'low risk' for Down Syndrome (approximately 20%). Noting this low risk, the physician did not inform the plaintiff-parents of the possibility that her fetus could have a genetic condition and did not offer further confirmatory testing. The pregnant mother eventually gave birth to a child with Down Syndrome, which she alleged was against their reproductive desires. In this case, the court held:

> A mere 20 percent chance (that the child would have Down's Syndrome) does not establish a 'reasonably probable causal connection' between defendants' negligent failure to provide the AFP test and plaintiffs' injuries. A less than 50-50 possibility that defendants' omission

> caused the harm does not meet the requisite reasonable medical probability test of proximate cause (212 Cal. App. 3d. 696 at 703).

In 1989, a 20% chance of disability was not seen as "reasonably probable causal connection" to hold physicians liable for a child being born with an 'unwanted' genetic variation (212 Cal. App. 3d. 696 at 700). However, as prenatal technologies begin detecting more conditions with greater precision, the courts' allowance of acceptable risk or error by providers decreases. In this way, the tolerance for those with disabilities or genetic differences also diminishes, as technological innovations sharpen the expectation that these conditions should be precluded with little room for error. Fast-forwarding to 2015, we see that plaintiffs in *Wuth ex rel. Kessler v. Lab. Corp. of America* (189 Wash. App. 660) were awarded damages amounting to $50 million for WBL claims because the defendants neglected the parents' explicit desire to avoid reproducing a child with a known and rare severe genetic condition. Here, the pregnant mother was able to pursue CVS prenatal diagnostic testing; however, because her clinicians failed understand the expecting couple's concerns and this did not provide her with appropriate genetic counseling services, the fetal condition was left undiagnosed, and the pregnancy was brought to term with a false reassurance of the to-be child's health. In *Wuth*, the court was far less tolerant of the defendant's errors, as contemporary medical knowledge and genetic technologies had more capacity to not only identify these possible outcomes but enable parents to prevent these births all together (e.g., via termination, alternate assisted reproductive approaches).

Comparing *Simmons* with *Wuth* reveals how tolerance for 'errors' decreases over time as physicians' professional scope is firmly expanded to include an informed use of genetic technologies. With this, acceptance of disability too narrows, as children with disabilities represent medical 'mistakes' that are sub-standard to contemporary biomedical expectations. Unlike *Simmons*, which involved Down Syndrome—a relatively common condition—the genetic condition pertinent in *Wuth* was exceptionally rare, requiring the use of a specifically sensitive FISH analysis.[30] In *Wuth,* the child Oliver was born with a translocation of his chromosomes 2 and 9; genetic material from these two chromosomes had been exchanged, causing his severe disabilities. This genetic translocation was known to Oliver's father, Brock, as he described his family having a history of birth defects with no known cause until his cousin with severe disabilities was genetically tested in 2003, when the technology to uncover her genetic variations was developed. However, because the physician misunderstood these genetic risks, conducted inadequate testing, and misinterpreted the results, simultaneously failing to rely on a genetic counselor, Oliver was born with the same translocation (189 Wash. App. 660). Given this clear provider negligence in light of contemporary technological expectations, care standards, and the severity of the condition, the plaintiffs' claims were ultimately successful. Through adjudicating such cases, we see that the courts' tolerance of error and risk in prenatal testing narrows as technological capacity and medical genomics knowledge is augmented, expanding the bounds around which disabilities represent legal injuries.

[30] Fluorescence in situ hybridization (FISH) is used to "visualize and map" genetic material "including specific genes or portions of genes ("Fluorescence" n.d.)." It is used for particularly complex cases and can "detect genetic abnormalities that include different characteristic gene fusions or the presence of an abnormal number of chromosomes" within a cell (Ratan et al. 2017).

As WBL involving prenatal technologies cases become more visible, courts also become conscious about protecting the medical institution from burdens of litigation. In 1979, *Berman v. Allan* (80 N.J. 421 at 432) was cautious to not "place too unreasonable a financial burden upon physicians." The court discussed the medical profession as one of "high esteem" within society, emphasizing that "physicians are the preservers of life" (80 N.J. 421 at 430). They tempered the damages and fault attributed to defendant-physicians by uplifting the societal respect owed toward medical professionals. Similarly, *Simmons* entirely rejected plaintiffs' damages, noting WBL cases "encourage costly and unreasonable over-testing and overtreatment for defensive purposes" because "physicians would find it necessary to place the requirements of the legal system before the needs and the finances of the patient." Concerns related to 'defensive medicine' are especially relevant in prenatal care, as ObGyns encounter the highest rate of malpractice claims (Guardado 2017, p. 8).[31] While physicians were still held accountable in WBL cases, courts were also invested in protecting the medical institution's social role to deter "medical malpractice premiums, resulting in an upward spiral of consumer costs," reflecting the way the appropriate use of prenatal genetic technologies are interpreted in a neoliberal privatized healthcare context (*Berman* 80 N.J. 421; *Simmons* 1989).

Over the years, prenatal testing has been constructed as a set of tools that allows pregnant individuals to make reproductive choices with as much health and genetic information as possible about their fetus, reassuring people about their pregnancies and allowing them increased control over the specific baby they will birth. Today, testing is not only routinized but celebrated as part of pregnancy medical management. For example, in 2007, ACOG recommended Down Syndrome screening for all pregnant people, regardless of age, emphasizing that the perceived advantages of early detection (i.e., enabling termination) makes widespread testing appealing (ACOG 2007ab). In 2011, the availability of NIPT marked a further push towards routinization of prenatal genetic screening. NIPT detects fetal genetic conditions via a standard blood draw from the pregnant person as early as 9 weeks in a pregnancy. Initially, NIPT was presented as a screening for those at higher risk for pregnancies with Down Syndrome, requiring subsequent amniocentesis to confirm any findings. Now, NIPT is implemented for *all* pregnant people, at times without pregnant people being fully cognizant that they have undergone genetic testing (discussed in more depth in following chapters). NIPT detects Down Syndrome with high sensitivity (99.9%) and specificity (98%). In a recent study conducted at 35 international centers, NIPT detected 100% of fetuses with Down Syndrome, while standard screening detected only 78.9% (Norton et al. 2015: 1593). Given its reliability and ease of implementation, NIPT represents a paradigm shift in prenatal genetic testing, eliminating the risk of miscarriage that invasive tests bear while offering reliable risk-based results for a variety of genetic conditions (Benn and Chapan 2009, p. 2154). Still, this technology, like its predecessors, has ramifications when it comes uncritical routinization that results in patient misinformation. In the larger picture, uncritical use of these tools can engender systemized pressures to de-select fetuses with genetic differences, narrow tolerance for disability, and undermine efforts to develop inclusive social structures.

[31] I attended several genomics research conferences as part of the broader methodology informing this dissertation. At one national conference in 2021, an insurance company representative noted that "defense against wrongful birth (lawsuits) is a big concern" when deciding which prenatal genetic tests the company will help financially cover for patients. Accordingly, we see how WBL suits continually go hand in hand with the routinization of (and increased covered access to) prenatal genetic technologies.

Routinization of prenatal genetic testing as a part of pregnancy management in the 1970s-1990s, and continued expansion of technological capacities since, has heightened expectations of testing implementation and accuracy while narrowing the tolerance for risk or error during the testing process. As a result, prenatal testing continues to pose substantial questions regarding disability justice and the potential elimination of certain disabled communities (Garland-Thompson 2015; Zhang 2020). This concern is especially salient when it comes to NIPT; this technology can not only yield false positives, but its results are *not diagnostic*, indicating only the probability that a fetus has chromosomal differences (Nuffield Council on Bioethics 2017). There are well-founded apprehensions that pregnancies will be terminated based on inaccurate, indefinite, or uncertain information from NIPT, with this decision potentially swayed by providers who may not thoroughly understand how to engage risk-based genomic data or provide equitable information about raising children with disabilities (Newson 2008, p. 103; Bayefsky and Berkman 2022; Kliff and Bhatia 2022; Meredith et al. 2023). As such, although NIPT, like other prenatal genetic technologies, can position patients as informed partners alongside their ObGyns and empower parents' reproductive choices, these tools need to be more thoughtfully implemented to mitigate possible systematic increases in abortions (specifically, medicalized and social pressures to terminate) due to fetal genetic differences and disabilities (Zhang 2020, Meredith et al. 2023).

The (Parental) Responsibility to Use Prenatal Genetic Technologies Toward 'Healthy' Births

While the discussion above illustrated how biomedicalization influences broader expectations around how reproductive technologies should be used, as enshrined in legal judgments, this section highlights the consequences of such technological routinization taking place within a neoliberal context – one which prioritizes individualism, embodied independence, and economic productivity. Innovation and routinization of genetic technologies within neoliberal societies gradually change ideas around who is responsible for using such tools toward specific ends. The WBL judgements reveal a steady 'technological imperative' placed on individuals (first providers, later parents) in that they *ought* to use available technologies to benefit themselves and broader societal outcomes (Rose 2001). This is perhaps unsurprising in the United States' privatized health context, where individuals are commonly held responsible for using technologies to surveil and ascertain their health and that of their family (Mukherjee et al. 2022). Women are expected to diligently follow reproductive cancer screening guidelines, men (especially those of color) are accountable for managing cardiovascular risks, and all individuals must be aware of their family medical history and mitigate risks from a relatively young age (Shim 2005; Rose 2001). This 'self-surveillance' responsibility requires people to 'sign on' to using technologies as an integrated aspect of managing their lives and moreover as an obligation to society (e.g., reducing public health burdens), much like what we see emerge in WBL cases involving prenatal genetic testing (Clarke et al. 2003; Rose 2001). As this discussion will show, technological imperatives are especially salient for pregnant women, as the gendered concept of 'good motherhood' extends to expecting women to use prenatal technologies to make 'responsible' choices about who they bring to life (i.e., children who conform to a vision of ableist health, normalcy, and functional socioeconomic productivity) (Denbow 2015; Ikemoto 1992; Rapp 1998; Rapp and Ginsburg 2001).

Shifts in who is responsible for using prenatal genetic technologies toward producing 'normal' 'productive' children are also intertwined with available reproductive options. When abortion is a safe and legal reproductive option, the (implicit or explicit) pressures expecting pregnant people to use prenatal technologies and make 'responsible' choices to terminate pregnancies with

disabilities or impairments can be amplified (Vecera 2014). To be sure, it is not access to abortion that is the issue – abortion should always be accessible to pregnant people for whatever reason they may need. There are a multitude of reasons that pregnant people seek abortions. Abortions are critical and life-saving maternal care when a pregnancy is unviable (e.g., ectopic pregnancies, miscarriages) or when the pregnancy, labor, and delivery process pose serious risks to the pregnant person's health and life. They can also spare a family and to-be child immense grief and suffering in cases where a fetus is affected with complex genetic anomalies or structural issues that would not lead to life or lead to the to-be child passing soon after birth. In other instances, a person may simply not want to be pregnant, or not be seeking to raise a child with additional needs as identified in utero (Glass and Meek 2023). Widespread, safe, and legal access to abortion represents fundamental healthcare. Rather than abortion itself, the issue here is how imperatives around prenatal genetic technologies are interpreted within a neoliberal context that holds a narrow perception of meaningful existence and prioritizes individually embodied independence over collectively beneficial societal inclusions. As the discussion unveils, technological developments become obstacles for social inclusion and equity when we (in this case, via courts) engrain pressures and incite accountability to use these technologies to systematically preclude births that don't represent neoliberal ableist ideals of 'normal' or meaningful, productive existence.

Reflecting neoliberal frameworks that privatize health responsibility, WBL case judgements individualize accountability for managing disabilities to parents, in their choice to test and/or terminate a pregnancy, or to defendant-physicians when testing is negligent. In particular, the degree of responsibility attributed to parents (versus physicians) shifts based on how courts interpret the appropriate use of prenatal genetic testing at a given time. While courts may reconceptualize liability in WBL cases, the notion that testing should be used to prevent genetic conditions and disabilities, and that supporting those disabilities is a private endeavor, is increasingly emphasized.

To understand how prenatal technologies 'should' be used when adjudicating WBL conflicts, courts rely partly on available guidance from professional medical societies. In the absence of such guidance, parents are largely responsible for their own reproductive outcomes. When existing professional guidance is sparse, as it is in relatively early cases, parental responsibility toward raising children with disabilities is emphasized, despite the availability of prenatal technologies (e.g., amniocentesis) that could have factored into parents' decision to bring the fetus to term. As discussed earlier, in *Park* (No. 190033, Cal. Super. Ct., Orange Cty., Dec. 13, 1974), amniocentesis (and prenatal diagnostic testing in general) was not within the contemporary standard of care; as a result, the defendant-physician could not be considered negligent for failing to test the pregnancy for Down Syndrome. However, the onus on parents shifts once expectations of prenatal genetic technologies and associated medical standards are established in professional statements. Professional guidelines first aim to routinize prenatal testing, and later focus on specific provider practices associated with various prenatal genetic technologies. This marks a change in the circumstances surrounding which actors – providers or parents – are responsible for the use of prenatal genetic technologies and potential consequences related to raising disabled children.

The court's reliance on professional medical guidelines released in the late 1970s and early 80s set the expectation that physicians should offer appropriate genetic tests and results during one's pregnancy or be held accountable for failing to do so. Recall, in 1979, the NIH stated that

amniocentesis should be routinely offered to women older than 35 years of age (National Institutes of Health 1979). This statement, along with other similar guidance from professional societies, including the AAP and ACOG, focused on routinizing prenatal genetic testing and marked a change in subsequent WBL judgments in terms of parental responsibility. In *Curlender v. Bio-Sciences Laboratories* (106 Cal. App. 3d 811 [1980]), the infant-plaintiff brought a WL action through her father, alleging that the defendant-testing laboratories where negligent in giving her parents "incorrect and inaccurate" information, thus representing the proximate cause of her birth and subsequent suffering. The lab had tested the infant's parents for Tay-Sachs genes and provided false negative results; this led to the pregnancy being carried to term and the infant-plaintiff being 'wrongfully' born with Tay-Sachs. The California Court of Appeal had "no difficulty in ascertaining and finding the existence of a duty owed by medical laboratories engaged in genetic testing to parents and their as yet unborn children" (106 Cal. App. 3d 811 at 828). Additionally, Justice Jefferson noted strong public policy considerations for recognition of this breach, given the advances in medical knowledge and genetic testing that represented the contemporary standard of care required to "prevent a genetic disaster" such as the Curlender family endured (Wright 1978, p. 1499; *Curlender* 106 Cal. App. 3d 811 at 827).

In the 1980 *Curlender* case, the court's dicta suggest that when current technology can reasonably promise an alternative to birthing a disabled child—and when the use of such technology is routinely expected as part of professional medical guidelines—parental responsibility is less emphasized in the face of physicians' or geneticists' wrongdoing. In this way, *Curlender* represents a significant expansion of California tort law, wherein the infant-plaintiff's cause of action was legally recognized, awarding damages for pain and suffering, and any special monetary loss resulting from Tay Sachs. In addition, her parents were awarded damages for their separate WB claim (106 Cal. App. 3d 811). This was the first time in California tort law that a WBL claim was not only recognized in favor of the plaintiffs, but that monetary damages where awarded.

Regarding *Curlender*, it is worth also noting that Tay-Sachs is a particularly severe and almost always lethal disease. Disease severity impacts the outcome of WBL cases in that plaintiffs are more likely to be seen as having suffered or experienced pain when the condition is life-compromising. Further, judges often do not perceive parents to be solely responsible for raising children with more severe conditions, justifying additional financial support through damages. This is perhaps also reflective of the individualized approach to managing health and disabilities in the United States, where private resources are emphasized over public infrastructure and supports. By comparison, in *Andalon v. Superior Court* (162 Cal. App. 3d 600 [1984]) the plaintiff-infant alleged that the defendant-physician was negligent in not offering his parents prenatal genetic diagnostic testing based on their risks and established professional guidelines, which caused him to be born with Down Syndrome, a chromosomal condition that is not as severe or life-threatening as Tay-Sachs is. Here, the court recognized that the parents and infant were "direct victims of the malpractice alleged"—the parents through the "burden of parental responsibility" and the infant due to his genetic condition—but did not award damages, positing the family still enjoyed togetherness despite the child's condition (162 Cal. App. 3d 600 at 611).

Cases following *Curlender* adopted a similar rationale regarding physicians' breach of duty. The *Turpin v. Sortini* (31 Cal. 3d 220 [1982]) and *Harbeson v. Parke-Davis* (656 P.2d 483 [1983]) courts separately found for the plaintiffs, recognizing the physicians' breaches of their duties to

appropriately inform, test for, and prenatally diagnose health conditions in the fetuses. Often, courts noted the value of informed consent in such cases: without prenatal genetic information, parents could not be considered fully informed in their reproductive decision-making. In this moment, accurate genetic information (via the appropriate use of prenatal genetic technologies) during pregnancy care thus becomes a necessary component to invoke parental responsibility. Parents cannot be entirely individually responsible for opting into prenatal genetic technologies and/or raising disabled children in cases where providers have not fulfilled their individual professional duties to offer prenatal genetic technologies (and accurate results) where appropriate.

Alongside professional guidance routinizing testing, the 1973 passage of *Roe v. Wade* (410 U.S. 113) significantly influenced how courts interpret parents' and providers' responsibilities surrounding prenatal genetic technologies in the late 1970s onward (Vecera 2014).[32] Prior to 1973, physicians were typically not held responsible for a child's congenital conditions, since—legally—nothing could have prevented the outcome, despite the expectation that prenatal testing technologies should be used. Before *Roe,* termination was both legally unjustifiable and morally questioned, as evidenced by *Gleitman* (227 A.2d 689) in 1967. Here, as discussed, the defendant-physician failed to advise the plaintiff-mother about the risks of rubella during pregnancy; but the judge noted the physician could not have prevented the birth, legally. The judge also adopted a moral tone, emphasizing the need to protect the 'preciousness' of a fetus: "it may have been easier for the mother and less expensive for the father to have terminated the life of their child while he was an embryo, but these alleged detriments cannot stand against the preciousness of the single human life to support a remedy in tort" (*Gleitman* 227 A.2d 689 at 693). However, with *Roe* enabling the choice to abort a fetus, courts had to take into account parents' right to *not* have a child, including when prenatal indications could determine "the child would be born deformed" (*Park* 60 A.D. 2d 80 at 88). In many ways, *Roe* was critical to shifting parental responsibility again – not only were physicians held accountable for ensuring parents could exercise their right to reproductive choice around birthing particular fetuses, but there also emerged an implicit understanding that expecting parents would want to (or should) terminate fetuses presenting with genetic conditions or disabilities. In WBL cases following *Roe*, we see how the framework of reproductive rights appears limited and once again places accountability for health and disability on individuals. One's reproductive 'choice' is enabled and constrained by neoliberal ableist ideals, and reproductive justice– the idea that equitable social structures should enable dignified parenthood for all, and equally celebrate the birth of disabled children– is fundamentally precluded (Luna and Luker 2013; Bowen 2022).

In the mid-late 1970s, *Roe* brought abortion within the scope of possible parental options and integrated it into the medical standards of prenatal care alongside routine prenatal testing (Vecera 2014). Given how reproductive autonomy is interpreted within WBL cases—to emphasize disabilities as unwanted—abortion is often reinforced as the preferable decision when prenatal testing reveals genetic anomalies (Hensel 2005, p. 177). Post-*Roe,* defendant-physicians were negligent and liable for a child's congenital conditions when they failed to inform parents of their right to abortion following prenatal genetic testing. Further, compensation in WBL cases becomes available only to parents who explicitly state they would have aborted their disabled fetus rather

[32] Given the United States Supreme Court's 2022 *Dobbs v. Jackson* judgement (Horn 2022), this relationship between technologies, abortion, and individual accountability would need to be reevaluated once more, particularly in states where abortion is now effectively illegal or prohibited after only a handful of gestational weeks.

than carried it to term. Parents unwilling to engage this claim are left to privately support their disabled children, without damages being awarded, and are often painted as "destructive to the family, socially irresponsible, and possibly immoral" (Asch 1989, p. 88).

In 1979, *Berman* (80 N.J. 421) enshrined these changes to parental responsibility post-*Roe*, serving as precedent for subsequent cases. The defendant-physician failed to offer amniocentesis to the 38-year-old plaintiff-mother, resulting in the child being born with Down Syndrome. Ultimately, the court awarded the plaintiff emotional damages:

> In failing to inform Mrs. Berman of the availability of amniocentesis, defendants directly deprived her — and, derivatively, her husband — of *the option to accept or reject a parental relationship with the child* and thus caused them to experience mental and emotional anguish upon their realization that they had given birth to a child afflicted with Down Syndrome. (80 N.J. 421 at 433, *emphasis added*)

The courts noted that the plaintiff-mother's constitutionally protected right to exercise her reproductive autonomy had been wrongfully denied as a result of the physician's negligence in failing to offer appropriate prenatal genetic testing. They emphasized that the plaintiff-mother had been robbed of "the opportunity to determine her destiny in whether or not to give birth to a gravely handicapped infant" (80 N.J. 421 at 436). Specifically, they recognized her loss of the chance to abort the disabled fetus; abortion was seen as a meaningful choice particularly when it involved preventing the births of children known to have congenital conditions. The court also cautioned about future implications of this case, noting that any other decision would "immunize" physicians from liability in providing guidance pertinent to individuals' constitutional reproductive rights (80 N.J. 421 at 432). Over the following decade, there was significant reliance on *Berman*, with heavy weight placed on the protected right to exercise reproductive autonomy in terminating disabled fetuses (we see this similar reasoning in both *Harbeson* 656 P. 2d 483 and *Simmons* 212 Cal. App. 3d. 696). In interpreting reproductive rights and prenatal genetic technologies within a neoliberal context, courts tended to focus on termination as a viable 'solution' for precluding the births of fetuses revealed to have genetic conditions per prenatal test results. Parenting a particular child—specifically one with disabilities—was no longer inevitable, according to the courts. Parents had more choice on *who* they wanted to raise, with courts and doctors playing a critical role in upholding and cementing values around celebrating normative health and ability. Regarding parental responsibility, *Roe v. Wade* (410 U.S. 113), alongside the court's reliance on professional guidance, now rendered this obligation unduly imposed where physicians failed to conduct testing or inform of the option to terminate a fetus with disabilities.

Around the 1990s and early 2000s, professional guidance began to specify the 'reasonable' practices physicians should undertake when implementing testing (partly to protect doctors from malpractice suits). Once again, courts leaned on these guidelines when responding to WBL conflicts between families and providers regarding the appropriate use of prenatal testing. However, the specificity of these new guidelines makes it more difficult for plaintiff-parents and plaintiff-infants to win their WBL claims and be awarded damages, as evidenced by *Johnson v. Superior Court* (101 Cal. App. 4th 869 [2002]) and *Barragan v. Lopez* (56 Cal. App. 4th 997 [2007]) WL cases where plaintiffs were unsuccessful. In *Johnson*, the plaintiffs alleged that their donor sperm was not properly tested for genetic conditions, passing down Autosomal Dominant Polycystic Kidney disease to the child (as the child is the plaintiff in WL cases, the parents did not

establish a desire to have otherwise terminated this pregnancy). The defendants, however, underscored that the affected gene was not their 'fault,' and that the child lived a sufficiently meaningful life even with the condition. That is, the child was not 'affected enough' to have warranted their life being precluded. In *Barragan*, while the plaintiffs asserted their physician failed to advise them of the right to termination which ultimately led to plaintiff-child being born with cerebral palsy, the physicians denied this duty given that there was no prenatal indication of the child's condition. In other words, they had used prenatal technologies appropriately, and seeing as there was no evidence that the to-be child would be disabled, there was no need to discuss termination. Here, we see how the right to termination, and the physician's duty to discuss this right, is only invoked when fetuses present with disabilities or genetic conditions.

Over time, there are also fewer WBL cases in California, with the most recent being in 2007. It is possible that advanced understanding of prenatal technological capacities (and known risks, e.g., false negatives) paired with increased specificity in professional statements about appropriate care creates a situation where physicians can protect their definition of the standard of care, and parents are more individually responsible for opting into testing and asserting their reproductive decisions, potentially deterring parents from pursuing costly WBL litigation. Still, preventing the birth of disabled children remains the focus in adjudicating individual responsibility in recent WBL cases.

From 1986, the California Health and Safety Code required clinicians to advise all pregnant patients of AFP testing, and by the early 1990s prenatal genetic testing was being routinely used in clinical settings. By the start of the 2000s, professional associations including ACOG and ACMG released numerous statements detailing parameters for appropriate clinical use of contemporaneous genetic testing technologies, ranging from diagnostic procedures like CVS and amniocentesis to carrier screening for specific genetic conditions including Fragile X, cystic fibrosis, and spinal muscular atrophy. With courts adjudication of WBL claims increasingly relying on these medical communities' more specific definitions of the standards of care, parents are increasingly responsible when their children are born with genetic conditions—unless they can prove provider negligence within these shrinking boundaries.

Subsequent to the 1990s, successful WBL cases necessitated unquestionable wrongdoing by physicians, as defined by their own professional parameters. In *Reed v. Campagnolo* (332 Md. 226 [1993]) and *Gami v. Mullikin Medical Center* (18 Cal. App. 4th 870 [1993]), the courts awarded damages to the plaintiffs, as the defendants failed to perform AFP testing and amniocentesis (both tests fell within well-established contemporary standards of care). Similarly, in the 2015 *Wuth* case (189 Wash. App. 660), the plaintiffs were not provided with a genetic counselor, and the physician failed to properly test, analyze, and counsel them on their prenatal testing results. The defendants were found liable because of unquestionably negligent prenatal care, unjustifiable given the contemporary professional guidance and technological expectations.

However, plaintiffs are not always successful based on asserting their right to abortion and pointing to lapses in physicians following professional societies' testing guidelines. Importantly, accuracy of prenatal genetic testing results is not guaranteed; there can be issues with false positives, false negatives, and findings for genetic screenings are almost always based on probabilistic risk calculations rather than definitive health outcomes (Bayefsky and Berkman 2022; Kliff and Bhatia 2022). *Barragan* (56 Cal. App. 4th 997) revealed that when technology fails within the acceptable

risk or error thresholds, parents are individually responsible for raising their disabled children. The plaintiff-mother gave birth to twins with cerebral palsy following prenatal testing results inaccurately indicating the fetuses did not have the condition. Still, the court ruled in favor of the defendant-physicians: "it is undisputed that, other than not advising mother of her right to an abortion, Dr. Lopez provided medical services within the standard of care" (56 Cal. App. 4th 997 at 1003). It was not considered necessary for the plaintiff to have been informed of her abortion right, because testing results indicated healthy fetuses; this further supports the argument that WBL claims are specific to precluding or compensating for *disabled* existence. When technology falls short within its acceptable contemporary parameters, parents bear responsibility toward caring for children with disabilities.

Parental responsibility has become more pronounced with time, as courts heed medical professional perspectives on technological advancement and bolster abortion as a recourse when prenatal testing indicates fetal anomalies. Given courts' consistent reliance on medical societies' guidance when resolving WBL conflicts, judgments skew to reflect medical communities' (rather than families') construction of appropriate clinical implementation of prenatal genetic technologies and uphold the medical model of disability. This is also seen in how courts shift to expressly protecting physicians and medical institutions from 'overzealous' malpractice litigation, as discussed. With increasing practice of defensive medicine, and courts interpreting prenatal genetic technology according to medicalized perspectives, the window for plaintiffs' success in WBL claims has gradually narrowed. Fewer practitioners are found negligent in recent WBL claims, suggesting parents have an individualized obligation to raise their (disabled) child even when they may have expected prenatal testing to preclude these births (Fox 2019). Aligned with the neoliberal ethos of privatized health management, courts' responses to decades of genomics experience, reproductive autonomy legislation, and more stringent clinical guidance have come to place responsibility of making judicious reproductive decisions about testing largely on parents.

Qualifying Disability and 'Meaningful' Life Alongside Technological Routinization

This section illustrates how the processes of biomedicalization, especially when situated in a neoliberal context prioritizing individualism and productivity, has lasting effects on how we collectively conceive of disability and meaningful existence. Over time, the routine reliance on prenatal genetic technologies to prevent 'impaired' existences while celebrating the birth of 'normal' children changes views on which lives are societally valued. How do we determine whose life and what types of existence are worth living? And how does the 'correct' use of technologies construct the 'normal' as delineated from the 'abnormal'? Courts contending with WBL cases navigate these very questions and concepts as they decide whether and how to compensate plaintiff-families with disabled or genetically affected children. As courts define disability as a legally cognizable injury, and seek to compensate families, they steadily reinforce the notion that some existences are 'normal' and worthwhile while others are 'abnormal' and undesired. Here, we also see how these boundaries between meaningful and unwanted existences are intertwined with ableist neoliberal ideals around embodied independence and economic productivity. Over time, as prenatal genetic technologies are positioned as a routine part of pregnancy care, the tolerance for diverse existences and genetic variations continues to narrow (discussed in the following chapter).

Once again, these changes also bear on reproductive politics, where one's reproductive rights are upheld and invoked particularly alongside technological imperatives to use testing toward

precluding disabled existence. While widespread access to abortion must be a staple of any equitable health system, when reproductive rights are interpreted in a narrow context stigmatizing disability, it jeopardizes the very principle of bodily autonomy and equitable reproductive choice. When certain individuals (from marginalized backgrounds) are excluded from parenting, disabled children are considered burdens on the State, or when disabilities and genetic conditions are considered legal harms and unwanted existences – as this discussion will illuminate – there is diminishing potential for meaningful reproductive and disability justice that can benefit society at large (Bailey and Peoples 2017; Luna and Luker 2013).

When adjudicating WBL cases, courts contend with the question of damages: should plaintiffs be awarded compensation for children born with disabilities, and do damages position disability as a legal injury? In engaging these issues, judges express their understandings of different existences and qualities of life. Over time, judges change how they conceptualize disability, though it is consistently framed as a 'tragic' and 'defective' outcome. Judges tend to frame disabilities as inherently unwanted and often espouse conventionally ableist values, reasoning "the mere fact of disability, without more, necessarily limits life's enjoyment" (Bagenstos & Schlange 2007, p. 755). In seeking to understand the 'injury' caused by disability in WBL cases, courts move from comparing the value of disabled existence to non-existence (that is, never having been born), and later comparing disabled existence to 'normal' existence. This shift, along with various nuances involved in judges' qualification of different existences, is shaped by legal definitions of reproductive autonomy, testing expectations, and health-related public policies. How disability is defined at a given time informs decisions and justifications surrounding damages awarded to plaintiffs, with courts ultimately recognizing disability as a legally cognizable injury worthy of compensation. We see how courts are consequential in co-producing how disabilities are conceptualized, and moreover regretted, as the 'proper' use of genetic testing innovations is interpreted in ways that steadily reinforce neoliberal expectations around preventing such reproductive outcomes and existences.

Early WBL cases frame life in itself as precious, worthwhile, and deserving of existence, despite contemporary prenatal technological capacity. In a 1967 WL case, the *Gleitman* court reasoned Jeffrey, who was born with compromised hearing, sight, speech, and several physical impairments, would still choose disabled life over no life at all:

> If Jeffrey could have been asked as to whether his life should be snuffed out before his full term of gestation could run its course, our felt intuition of human nature tells us he would almost surely choose life with defects as against no life at all. (227 A.2d 689 at 697)

The courts described Jeffrey's existence as "defective" but noted "a child need not be perfect to have a worthwhile life." While his disabilities were seen as representing a less than whole existence, his existence overall was interpreted as meaningful to some extent (at least meaningful enough in that it was preferred to non-existence). Accordingly, the court emphasized the Gleitman's parental responsibilities toward caring for Jeffrey, stating that "the right of their child to live is greater than and precludes [their parental] right not to endure emotional and financial injury" for their disabled child (227 A.2d 689 at 692). Here, it is important to note that prenatal genetic testing was nascent at this time and alternatives to birth were limited in this pre-*Roe* era, leaving the few other 'remedies' to Jeffrey's birth.

In subsequent years, however, there is a notable turn in how courts contend with questions around the right to life and disability. The early 1970s saw several precipitous moments attempting to define 'life.' Most notably, *Roe v. Wade* (410 U.S. 113) allowed for more liberal access to abortions nationwide. California also changed its state laws to hold fetuses as distinct from 'human beings' in homicide statutes, while leaving the term 'fetus' itself undefined (California Penal Code 187). These legal precedents came at a time when prenatal genetic testing and in-utero diagnoses, namely via amniocentesis and genetic carrier screening, were being introduced and quickly integrated into pregnancy care. Consequently, courts adjudicating WBL cases became more amenable to considering differing qualities of existence, drawing more distinctions between "normal" and disabled existence based on increasingly available prenatal genetic insights. While the value of life was still generally upheld, courts began recognizing disabilities as detracting from one's existence, and this was now being reflected in damages awarded to plaintiff-families.

Shortly after *Roe,* WBL judgments reasoned that framing a plaintiff-child's disability as a legal injury would suggest that the child should rather have not existed at all. While still describing disabilities as inherently unwanted, these judgments noted that life, in any form, is always more valuable than non-life—and thus, disability as an outcome cannot be a legal injury (when compared to non-existence in this way). However, given *Roe*, courts recognized reproductive rights as a critical part of the pregnancy care standard of the time. We see this distinction unfold in the 1979 *Berman* (80 N.J. 421) court, which awarded damages to plaintiff-parents in their WB case (for the deprivation of their right to termination, had prenatal genetic testing been conducted 'appropriately'), but denied the plaintiff-child's WL claims focused on seeking damages based on her disabled existence. The *Berman* court carefully framed their decision with regard to Sharon's (plaintiff-child's) disabled existence. Here, the doctors had failed to perform an amniocentesis while Sharon's mother was pregnant (at the age of 38, which is considered a 'high risk' for causing fetal chromosomal variations), thus neglecting to inform the plaintiff-mother that Sharon would have a much higher chance of being born with Down Syndrome. When reasoning through Sharon's WL claim, the courts described her as a "mongoloid child" with a "defective" and "tragic" existence. They defined her quality of life as "more circumscribed than those of normal, healthy children" and noted that she "will experience a great deal of physical and emotional pain and anguish." Rather expressly, they framed Sharon's disabled existence as less desirable than 'normal' existence. However, despite their pejorative conception of her disabilities, Sharon's existence was considered more valuable and enjoyable than the alternative of non-existence:

> Sharon, by virtue of her birth, will be able to love and be loved and to experience happiness and pleasure — emotions which are truly the essence of life and which are far more valuable than the suffering she may endure. To rule otherwise would require us to disavow the basic assumption upon which our society is based. This we cannot do. (80 N.J. 421 at 430)

Although Sharon's disability was contrasted with "normal, healthy" children, there was an understanding that because life always equates to some form of joy and pleasure, it must be preserved in all its various forms. To defend this rationale, the court invoked sovereign principles embodied within the US Constitution and Declaration of Independence, noting that life is "one of three fundamental rights of which no man can be deprived without due process of law" and every person has an "unalienable right to life is a self-evident truth." They stated, "one of the most deeply held beliefs of our society is that life — whether experienced with or without a major physical

handicap — is more precious than non-life." As such, we see how early WBL courts compared disabled life to non-existence when adjudicating whether and how damages should be awarded, and whether disability can be considered a legal injury. While Sharon's Down Syndrome was taken as a tragic outcome and the courts recognized a failure on the doctor's part to perform an amniocentesis, her WL claim was denied, and WB damages awarded to her parents were limited to emotional anguish. Damages associated with the costs of raising Sharon were denied, as the court believed that the family experienced sufficient love and joy despite her disabilities, and thus ought to provide for her needs (80 N.J. 421 at 429).

However, soon after, courts stop comparing disability to non-existence, and instead shift to comparing disability to their conception of 'normal' existence. In doing so, courts assert that while disabled existence is valuable as human life, the 'wrongdoing' causing the plaintiff-child's disability amounts to a legal injury, through deprivation of the child's 'normal' life. To assess the degree to which disability deviates from their conception of normalcy, courts evaluate a child's "functional limitations and the extent of [their] suffering in light of current medical knowledge and its ability to 'cure' such ailments" (Hensel 2005, p. 182). With genetic insights expected to illuminate more prenatal anomalies informing reproductive decisions, courts reflect and co-construct an expanding classification of disability. As they expand their understanding of which existences represent disabled life (simultaneously narrowing their conception of 'normal' life), courts begin including conditions not previously considered legal injuries, like Down Syndrome and deafness, into their classification of disabled life. In this process, courts emphasize bodily independence and physiological functions when conceptualizing disability, pointing to individualized medical interventions (rather than structural inclusions) as suitable remedies to one's additional needs. Importantly, plaintiffs must be willing to treat disability in a similar manner; they must frame their child's existence as an inherently unwanted or burdensome tragedy they would have rather avoided and individualize blame to medical providers or institutions in order to receive compensation (Hensel 2005, p.171). In this way, courts strengthen a medicalized understanding of disability as objective and embodied, rather than contingent on sociocultural or relational influences. This understanding of disability partly reflects increasing societal acceptance of prenatal genetic testing alongside the right to termination (when prenatal insights indicate genetic conditions or disabilities), and more knowledge about genetic causes (and treatments) for various congenital conditions (Vecera 2014).

This development of comparing disabled life to 'normal' life is demonstrated in the 1980 *Curlender* (106 Cal. App. 3d 811) case. The plaintiff-child Shauna (described as "genetically defective") suffered the "catastrophic result" of physician negligence, as she was "wrongfully" born with Tay-Sachs and would survive only for a few years. Here, the court dismissed the consideration of whether "the plaintiff might not have come into existence at all," noting WL cases relate to whether "a plaintiff *exists and suffers,* due to the negligence of others," and not whether they should have existed in the first place. Taking genomics to be well-established contemporary medical knowledge, they further stated "the certainty of genetic impairment is no longer a mystery" when underscoring defendants' negligence. The *Curlender* court established the "*right* of such child to recover damages for the pain and suffering to be endured during the (child's) limited life span (*emphasis added*)" (106 Cal. App. 3d 811 at 831). Awarding damages to plaintiffs, *Curlender* set into motion a paradigm shift wherein disability was seen as legally wrongful (as it

deprived the child of 'normal' life), justifying compensation for parents and children based on their 'non-normal' differences (106 Cal. App. 3d 811 at 815).

Once courts established that the 'injury' of disability ought to be compared against 'normal' existence, disability is seen not just as 'defective,' but at times courts establish that non-life may be preferable. In 1982, the *Turpin* court crystallized this treatment of quality of existence, disability, and damages for subsequent WBL cases:

> Considering the short life span of many of these children [with genetic conditions] and their frequently very limited ability to perceive or enjoy the benefits of life, *we cannot assert with confidence that in every situation there would be a societal consensus that life is preferable to never having been born at all.* […] Moreover, while our society and our legal system unquestionably place the highest value on all human life, *we do not think* that it is accurate to suggest that this state's public policy establishes — as a matter of law — *that under all circumstances 'impaired life' is 'preferable' to 'nonlife*.' (31 Cal. 3d 220 at 234, *emphasis added*).

In *Turpin,* Joy, the plaintiff-child, was born deaf, prompting the courts to consider the nuances of various disabilities and the extent to which they differently compromise the quality of life. Here, they contended with more abstract definitions of life, existence, and the human condition, particularly leaning on the 1976 Health and Safety Code which acknowledged the relative value of various human existences. *Turpin* characterized human life as having a "physical existence with the capacity both to receive and give love and pleasure as well as to experience pain and suffering," while understanding Joy's deafness as a severely limiting condition (31 Cal. 3d 220 at 237). As noted above, the court established that children with genetic conditions and congenital disabilities often had "very limited ability to perceive or enjoy the benefits of life" (31 Cal. 3d 220 at 234). This subjective value placed on human existence, and the balance of pleasure and pain as natural to the human condition, was crucial when comparing disability to 'normalcy' in future cases. *Johnson* used this classification in denying the plaintiffs damages (whose child faced a less severe disability treatable throughout her relatively long life), citing the child's experience of love alongside pain equated to a typical ('normal') life experience (*Johnson* 101 Cal. App. 4th 869 at 869). When compared to the courts' view of 'normalcy,' disability moves beyond physiologically embodied functional limitations, and is considered as detracting from the ability to enjoy the wholeness of life, at times inferior and less complete as an overall existence (Bagenstos & Schlange 2007, p. 749).

Wuth (2015) further developed this definition of desirable 'normal' life. The *Wuth* defendants relied on *Turpin* to absolve themselves, arguing that the plaintiffs experienced joy alongside their pain of having a disabled child. However, the jury found a "net loss" for parents, given the severity of the child Oliver's condition. This case also illuminated how parents must be willing to showcase their child's disability as a spectacle of less meaningful existence in order to win their WBL cases. In the hearings, Oliver's parents, Brock and Rhea, testified that their son looked "vacant" and "broken" at birth. They described how he did not look physically proportional, detailing the 'mismatched' size of his limbs and appendages. Further, underscoring a 'perverse' embodiment, they noted that Oliver had "inverted nipples and a buried penis" and that his "head was bent and turned," along with several issues in his muscles, tendons, and limbs. Brock and Rhea also emphasized Oliver's lack of normative development, stating that he did not have typical language,

vision, judgement, and fine motor skills, with his speech "limited to a few dozen words understandable only to his immediate family" (189 Wash. App. 660, 667-68). Ultimately, the court awarded the plaintiff family with damages totaling $50 million, strengthening the notion that disabled existence can be comparatively and objectively worse than other forms of existence (189 Wash. App. 660 at 841). When WBL courts consistently frame disability as inherently less positive and worthy of compensation they suggest that, while human existence involves pleasure alongside suffering, disabled life can represent a less meaningful, unwanted existence.

Perhaps the most significant precedent set by *Turpin* was modeling an approach for awarding damages, as it distinguished between special and general damages when understanding disability as an injury. *Turpin* reasoned that defendants had "interfered with the child's basic *right* to be free from physical injury caused by the negligence of others" (31 Cal. 3d 220 at 232). By failing to use prenatal genetic technologies 'appropriately' to prevent disabled births, WBL defendants had deprived the child of their right to be born 'normal' (or their parents' right to have not birthed the child at all). Henceforth, disabilities constituted a legal injury because the plaintiff-child "never had a chance 'to be born as *a whole, functional human being* without total deafness'; if defendants had performed their jobs *properly*, she would not have been born with hearing intact, but — according to the complaint — would not have been born at all," which would have been preferred by plaintiff-parents (31 Cal. 3d 220 at 233, *emphasis added*). The court held that a person *deserves* not to be born disabled, with disabled life representing a less than a 'whole' human being, and they further emphasized parents' right to choice regarding which children they want to bring to life:

> As the wrongful birth decisions recognize, when a doctor or other medical care provider negligently fails to diagnose a hereditary problem, parents are deprived of the opportunity to make an informed and meaningful decision whether to conceive and bear a handicapped child. (31 Cal. 3d 220 at 233)

Based on this reasoning, the court contemplated a model for awarding plaintiffs damages without framing disabilities as objectively 'bad,' while allowing monetary support for the material burdens associated with the 'tragedy' of disability. Like *Curlender*, *Turpin* compared disabled life to non-disabled life; however, unlike prior cases, *Turpin* established damages can and *should* be calculated in WBL cases, similar to other torts. The court rejected the idea that damages "would 'disavow' the sanctity and value of less-than-perfect human life," which had been commonly held in prior WBL cases. In doing so, they distinguished that damages do not compensate for life itself, but rather for the additional toll involved in disability. While general damages could not be accurately computed (given the inability to measure a counterfactual to life, i.e., non-existence), special damages were justifiable and measurable to support plaintiffs with "extraordinary expenses" and "medical expenses" required for their child's disability and additional needs (31 Cal. 3d 220 at 235).

This focus on awarding damages as something one private actor (e.g., defendant) owes another (e.g., plaintiff) resonates with the United States' neoliberal approach to privatized health management and lack of social supports for childcare and disabilities; here, raising children with diverse needs is the family's responsibility rather than the State's, with negligent defendants accountable for 'injuries' caused. Accordingly, in the United States, where caring for those with disabilities is framed as a private responsibility, seeking damages through the legal system is often one of the most substantial approaches for parents to support expenses related to their disabled

children's additional needs. Thus, in WBL cases following *Turpin* (1982), rejecting general damages while awarding special damages became a convenient arrangement for courts to ensure families can privately manage their child-rearing needs. In 1983, *Harbeson* (where infant twins were born with fetal hydantoin syndrome) cemented this approach, allowing parents to recover damages for "those expenses in excess of the cost of the birth and rearing of two normal children" and representing "extraordinary expenses for medical care and special training" for the children (*Harbeson* 656 P. 2d 483 at 483). In 1993, *Gami* also relied on this precedent in awarding special damages, further highlighting the growing bounds around disablement as it becomes considered a legal injury (18 Cal. App. 4th 870 at 870).

Other approaches to damages were unsuccessful. The *Andalon* plaintiffs tried to recover damages for "lost earning capacity" of their child born with Down Syndrome. Ironically, using *Turpin* as a precedent, the court stated that "one cannot lose what one never had" in terms of earnings and thus rejected the claim (162 Cal. App. 3d 600 at 600). Still, given subsequent cases where plaintiffs have won damages based on courts comparing disabled existence to normal existence, *Turpin* has strongly justified an approach to awarding special damages to attenuate the privatized emotional and economic tolls associated with disability in the United States.

There are also instances where courts are unwilling to contend with the depth of questions around disability, quality of life, and existence, which reveal some of the larger questions around justice and classifying meaningful existence illuminated in WBL judgements. Although, these judgements tend to precede the major shifts in how disabled life was conceived (in comparison to 'normal' life rather than non-existence) and the paradigms established to award special damages, which began to unfold in the 1980s. Unwilling to qualify life and existence, in *Becker* (1978), the court stated:

> Whether it is better never to have been born at all than to have been born with even gross deficiencies is a mystery more properly to be left to the philosophers and the theologians. Surely the law can assert no competence to resolve the issue, particularly in view of the very nearly uniform high value which the law and mankind has placed on human life, rather than its absence (46 N.Y.2d 401 at 411).

Becker viewed these questions around life and existence as transcending the role of the legal system and asserted the fundamental value of life itself. While they framed disabilities as deficiencies, they also problematized the "staggering" implications of finding disability a legal injury: "Would claims be honored, assuming the breach of an identifiable duty, for less than a perfect birth? And by what standard or by whom would perfection be defined?" (46 N.Y.2d 401 at 411). While the court raises important concerns – and presents critical questions palpable in each WBL case presented here – their refusal to contend with such issues exacerbates challenges faced by parents of disabled children in a neoliberal state. Scholars have argued that dismissal of WBL claims absolves physicians from their professional responsibilities toward parents, simultaneously emphasizing parents' individualized burden as they become solely responsible for supporting their child's additional needs in a privatized health system (Fox 2019). Within a context that understands meaningful existence per ableist neoliberal terms, courts are perhaps compelled to contend with such questions around defining disability, if only to afford just outcomes for parents of disabled children who have few other private resources. In the absence of robust inclusive social structures, where disabilities are seen as tragic private burdens, tort law approaches like those taken in WBL

cases emerge as *the* way for families to ensure they can individually support their disabled children's needs.

Where courts before the mid-1970s considered all life to be valuable (compared to non-life), changes around reproductive rights, public policy, and technological expectations shifted their perspective to ultimately claim that disabled existence is not always preferable. Eventually, judgments and damages clarify that disability can be 'worse' than not only normal existence but non-life as well, defining disability as tragic, defective, and a burden that should be compensated. Accordingly, over time, with heightened expectations around the breadth and accuracy of fetal genetic insights, and sharper focus on parents' reproductive choice to raise a particular child, more children are considered disabled in their deviance from medicalized understandings normative health and ability. This shift to defining disability in contrast to an increasingly narrow understanding of normative existence sharpens the boundaries around which lives are considered meaningful, with disability firmly representing a legally cognizable injury and an unwanted existence. Moreover, over time, children are seen as having the 'right' to be born without disabilities given routinized prenatal genetic insights, and providers are considered at fault when depriving families of this right through 'improper' use of prenatal genetic technologies. As disabilities are constructed as unwanted harms, with parents also having to disavow their child's existence accordingly, plaintiffs are increasingly awarded damages to compensate for disablement. Together, these changes in WBL judgements alongside technological developments underscore not only that disability is an objectively undesirable existence but that the responsibility for caring for those with different needs belongs to individuals rather than the State's public infrastructure.

Final Thoughts on How (Uncritical) Routinization of Prenatal Genetic Technologies Shapes Reproduction and Disability

This chapter illustrates the consequences of biomedicalizing reproduction toward neoliberal ideals, as prenatal genetic technologies become increasingly integrated into the routine practices and expectations surrounding pregnancy care and reproductive decision-making. Examining the adjudication of WBL cases over the past six decades shows how prenatal genetic technologies gradually become the crux of reproduction, amplifying expectations to not only utilize testing regularly but to do so toward the objective of reproducing normatively able-bodied children. As WBL judgements reveal, the interpretation and routinization of prenatal genetic technologies within a neoliberal context not only construes disability as an unwanted, unproductive embodied existence but also contributes to privatizing the responsibility of caring for those with diverse needs. Understanding how courts—as institutions that reflect and co-produce sociocultural values—contend with and reinforce these biomedical transformations provides insight into broader societal deliberations around the 'appropriate' use of prenatal genetic technologies, especially as this pertains to conceptualizing healthiness, disability, and meaningful existence. As courts decide on the 'proper' use of genetic technologies, they gradually reinforce the imperative to use such tools toward preventing the births of those who are dis- or differently abled, casting these existences as less fulfilling and societally unproductive.

Courts lean on the promises of prenatal genomic innovations, legal and policy precedents regarding reproductive rights, and guidelines from professional medical societies to gradually shape and respond to societal expectations of healthiness, privatized health responsibilities, and understandings of disability. First, health, including acceptable degrees of risk and error tolerance

when using genetic tools, is reimagined in continuous relation to developing technological capacities. As prenatal genetic technologies are enhanced to be more specific, accurate, and provide access to more fetal health information, there an increasing expectation that these tools will be used routinely during pregnancy management alongside a decreasing acceptability of errors leading to "genetic disasters" (106 Cal. App. 3d 811). Second, WBL judgments reinforce a privatized approach to health, where individuals (rather than states) are responsible for health management and disability supports. Parents are responsible for utilizing technologies and exercising reproductive autonomy (i.e., terminating fetuses presenting with genetic conditions), while clinicians must maintain the standard of care around using prenatal genetic technologies and providing accurate information that enables parents to privately manage their family's health. In both instances, health responsibility is privatized, and the overall objective remains precluding the birth of disabled children. Finally, while WBL judgements generally considered all life to be precious, over time some disabled children (based on the condition and the degree of provider negligence) are framed as "defective," embodying incomplete, unfulfilling existences. Parents, too, reify notions of disability as embodied and unwanted, presenting testimonies that lament their children's conditions and bodies as 'perverse' and tragic. Gradually, as prenatal technologies are routinized and individual responsibility toward producing 'normal' children is heightened, using these tools to prevent the birth of disabled babies is celebrated, and lapses in technological implementation resulting in disability increasingly amount to legally cognizable injuries (with damages awarded). Taken together, the emphasized technological imperative to test alongside the conceptualization of disability as inherently inferior and detractive from reproducing (economically productive) 'functional' human beings echoes neoliberal discourse surrounding how biotechnology and private health responsibility ought to create normatively able societies.

The emphasis on individual health responsibility goes hand in hand with genetic innovations in neoliberal contexts that undoubtedly shape our relationship with biotechnologies, health, and notions of valuable existence. Particularly, we see how biomedicalization unfolds as technological transformations are situated in a neoliberal setting, wherein healthcare is privatized, and families are individually responsible for children's health and disability needs. While genetic technologies are presented as a 'choice' to empower one's decision-making (and in this case, reproductive rights), this is often more of a pressure than it is an optional decision, engendering an "era of genetic responsibilization" (Reuter 2007; Knight and Miller 2021). This pressure is further amplified and gendered for pregnant women, as the imperative to use reproductive genetic technologies comes to define 'good motherhood.' Individuals (especially women) are held responsible for being active partners alongside their physicians and cooperating in the medicalized effort to preclude the births of children with genetic conditions. In this way, pregnant people are often (even implicitly) compelled to use testing rather than exercise their agency to use or decline these tools as informed decisions based on their personal values. 'Choice' in this case is more so a façade, with the 'responsible' decision framed (and moralized) as opting into using genetic technologies toward reproducing children who embody functional independence and can be 'productive' members of society. This idealization of embodied independence and productivity also reinforces reductive notions of genetic essentialism and biological determinism, wherein one's life outcomes are narrowly constrained to their bodily existence. The duty toward reproducing 'normal' 'able' children thus becomes one's individualized obligation toward society, achievable only via pursuing genetic testing. Importantly, framing genetic testing as an individual obligation absolves the State from fostering inclusive social structures that are beneficial to everyone, not

least because they enable those with diverse needs to realize their potentials and participate in society. (Reuter 2007; Rose 2001; Clarke et al. 2003; Denbow 2015; Rapp 1998; Apple 1995).

To be sure, it is not that prenatal genetic technologies are in themselves deleterious to individual and social wellbeing. These technologies can be important tools in empowering individuals' reproductive decisions, increasing understanding of health needs, and enabling timely medical care and interventions. When these innovations are implemented critically, foremost with ideals of social equity in mind, they certainly can represent useful biotechnological advancements. However, what this chapter, and those following, seek to illuminate are the harmful consequences that can take place when these reproductive genetic technologies are *not* implemented thoughtfully, and are rapidly routinized without consideration for the implications on reproductive justice, disability inclusion, and equitable values around patient-centered care. It is critical to realize the impact that reproductive genetic technologies (e.g., preimplantation genetic testing on embryos, prenatal genetic testing on fetuses, preconception genetic testing for potential parents or gamete donors) have on shaping our tolerance for diverse existences and heightening pressures to preclude certain births (Karpin and Mykitiuk 2021). It is even more essential to have these discussions in the current moment, wherein technologies like non-invasive prenatal testing (NIPT) are making it easier than ever to routinize genetic testing into reproductive care. Often covered by insurances and encouraged by public health programs, genetic technologies such as NIPT are easier to access (and easier to conduct, via blood draw) than their predecessors. As such, it is especially necessary to be critical of how these genetic technologies are being routinized into our reproductive practices, public health programs, and health laws and policies.

As NIPT becomes part of routine prenatal care, more fetal genetic differences will be discovered in-utero, with this information increasing in accuracy over time. Simultaneously, more people will be confronted with the potential decision to terminate pregnancies upon learning genetic insights. Currently, almost 70% of pregnant people in the United States abort fetuses identified to have Down Syndrome, despite the relatively promising prognoses attached to this diagnosis today (Natoli et al. 2012). Again, the issue here is not that people are aborting fetuses – the right to termination, for whatever reason, remains paramount to equitable healthcare. Rather, the problem emerges when fetuses with genetic variations and disabilities are being systematically de-selected because their existences are seen as unworthwhile or due to a lack of public social support for those with disabilities (thus placing an unmanageable private burden on families) (Zhang 2020). The largest current study of parents (242 respondents) who received a prenatal diagnosis of Down Syndrome between 2016-2021, well into the era of NIPT routinization, revealed that expecting parents regularly receive biased recommendations from their clinicians regarding raising a child with this condition. Following prenatal genetic tests that indicated a fetus had Down Syndrome, 61% of ObGyns presented results as a tragic outcome. Less than 40% of ObGyns discussed disability supports, services, and balanced life outcomes for those with Down Syndrome, with 64% of patient consultations focusing mostly on medical issues children may face and 76% most concerned with ensuring pregnant parents knew their reproductive options. Moreover, parents recalled their providers describing their child's condition as a "mistake" of nature, and being asked explicitly biased questions such as, "How do you feel about your child not having good quality of life?" or "How does it make you feel to know that you will die and leave your baby dependent on the state?" Given these systemic provider biases, one can imagine NIPT being uncritically used to influence individuals' reproductive decision-making, preclude and undermine the births disabled

children, and thereby exacerbate stigma toward disabled communities and neglect toward investing in robust structural inclusions. (Meredith et al. 2023)

It is worth noting that provider biases advising expecting parents to select against fetuses with disabilities often misrepresent the quality of life for disabled people and misinform parents that their child will not fulfill their expectations for joy and fulfillment in a family (Asch 2002; Knight and Miller 2021). Research shows that parents who receive balanced and thorough information about the resources and social supports available for their disabled child have more positive experiences within their families and with their medical providers (Lippman and Wilfond 1992). Rather than facing "agonizing experiences," families with disabled children, on average, report similar outcomes to families overall (Parens and Asch 1999). Although families do experience challenges when raising children with disabilities—especially in the face of structural barriers, lacking social services, and limited private resources—they still "flourish and show no difference in parents' stress levels, family functioning, and marital satisfaction" (Asch 2002; Mukherjee and Shirinian 2022). Biases against those with disabilities and the focus on ensuring 'genetic perfection' wrongly root one's quality of life in biological composition rather than social circumstances. While genetic conditions and disabilities are not inconsequential in shaping one's life, research underscores that factors like access to stable and affordable housing, education, food, employment, and healthcare are far more predictive of one's wellbeing and life quality (Parens and Asch 1999; Mukherjee and Shirinian 2022). Accordingly, as prenatal genetic technologies such as NIPT become more accessible and standardized in clinical care, it is essential that providers are better trained in taking a non-judgmental approach when presenting testing as a voluntary choice to pregnant patients. Providers must also consistently provide balanced, accurate, and non-discriminatory information around raising children with disabilities, being abreast of relevant social supports and resources that parents can harness. It is critical that the decision to continue or terminate pregnancies based on prenatal genetic findings lie with prospective parents, and that their providers make them aware of not only their right to abortion, but also relevant social supports that can facilitate raising a disabled child (Meredith et al. 2023; Knight and Miller 2021).

Finally, perhaps most salient in this current moment is the question around abortion in the United States. Despite the rapid expansion in prenatal genetic testing, access to abortion (and state support for those with disabilities) continues to shrink. In 2021 and 2022, abortion rights suffered tremendous blows across the country, with the Supreme Court overturning *Roe v. Wade (*410 U.S. 113) in their 2022 *Dobbs v. Jackson Women's Health Organization* ruling. Without the federally protected right to abortion, numerous states have banned or severely limited access to pregnancy termination. As a result, pregnant people, people of reproductive age, and their families across the country have suffered. Many cannot access life-saving abortions, abortions to prevent the suffering of fetus' with severe genetic anomalies or structural malformations, or must spend large sums of private resources (in some cases over $15,000) to travel out-of-state for abortions which are uninsured. Pregnant people in states where abortion is illegal or extremely limited have also largely lost access to general prenatal and reproductive care, as ObGyn and maternal fetal medicine (MFM) providers leave their practices. In these places, ObGyns and MFMs have expressed an inability to perform their jobs appropriately because they are unable to provide abortions as medically necessary for fear of legal retribution. Consequently, access to reproductive healthcare (including prenatal and post-pregnancy care) has declined significantly across the United States, magnifying the existing maternal mortality crisis nationwide (Glass and Meek 2023).

These waning freedoms especially impact marginalized communities, where individuals cannot afford to access safe and legal terminations through private means and are further entrenched in socioeconomic hardships when forced to bring pregnancies to term (Sawhill 2022). Further, more disabled pregnant people will suffer as a result of ending *Roe*, given how the medical system deprioritizes preserving the health and lives of those who are disabled, paired with the higher risk for pregnancy complications and maternal fatality for those who are disabled or have chronic conditions (Bowen 2022). Specific to prenatal testing, 13 states already prohibit aborting fetuses based on in-utero findings of disability. Some states also legislate limits on the number of abortions one can have following prenatal genetic testing, while others legally permit healthcare providers to withhold information about a fetus' genetic condition from parents (Bayefsky and Berkman 2022). As the United States contends with a future sans *Roe*, the relationship between reproductive genetic technologies, disability and health, and the responsibility for supporting diverse existences stand in the spotlight of lasting social transformations.

Given the intersection of prenatal genetic testing and abortion, and the short timeframe between genetic findings and abortion, these legal changes significantly impact one's reproductive options (Vecera 2014). Where abortion is not an option, the private sphere (including families and the private sector) necessarily assume more responsibility in health landscapes like the United States, where there are few inclusive structural remedies and public supports (Rapp and Ginsburg 2001, p. 397-8). Even in contexts where abortion remains safe and legal (e.g., California, Colorado, Oregon), we must be critical of how reproductive genetic technologies like NIPT are implemented and routinized. While these tools bear the promise of greater reproductive autonomy and useful health information, they can also amount to undue pressures and "intensified manipulation" that urge pregnant people to terminate certain fetuses and undermine disabled existence (Knight and Miller 2021). These tensions in how abortion and disability interrelate raise questions around balancing reproductive autonomy with reproductive justice. That is, while the right to abortion remains essential, it must be paired with a justice-based framework that dignifies the parenthood of all individuals and celebrates all births equally, fostering access to equitable public supports that are ultimately beneficial for society at large (Luna and Luker 2013). When it comes to prenatal genetic testing, being able to harness technological innovations responsibly and thoughtfully, foremost upholding values around social equity as intertwined with individual wellbeing, is integral to realizing the benefits these tools can bring.

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CHAPTER THREE

Of Boys and Girls:
Sex Chromosome Aneuploidies and their Diagnostic Implications

There is a quintessential moment in pregnancy when a doctor asks expecting parents whether they want to know the sex of their to-be child. Based on their response, the doctor will either reveal this information during a routine 20-week ultrasound or wait until the baby is born. In each instance, the doctor is looking for physical signs of a penis, at least 0.6 inches long, before declaring "Congratulations, it's a boy!" Or, in the absence of an adequate phallus, a girl (Fausto-Sterling 1993; 2013: 222). This declaration of 'boy' or 'girl' begins an individual's gendered existence. It sets expectations and rules for how that individual is to perceive themselves but more importantly how they are to be perceived by their family, friends, and society (West and Zimmerman 1987). In this critical moment, medical professionals map our physiological features (e.g., phallic size) onto our social identities (i.e., gender of boy/girl man/woman), so as to claim that the body provides 'truth' or an objective biological foundation that *determines* one's lived social experiences. Even though the penis and clitoris come from the same set of structures, and ovaries and testes arise from the same "indifferent fetal gonad," the medical institution has historically insisted that there is something fundamentally different about male and female genitalia on a sociocultural level, that our genitalia govern how we exist as gendered beings (Fausto-Sterling 2013: 219-220).

Gender is certainly not the only social category to show up in medical claims about how physical bodies objectively determine identities and experiences. Ideas around race and class have long informed medical pathology. Doctors routinely attribute poor health (e.g. cardiovascular conditions, obesity, diabetes) to one's racial ancestry with no consideration of structural barriers to health (Roberts 2011; Shim 2005). Pharmaceutical companies create drugs to target supposedly essentialized conditions in Black individuals (e.g., BiDil for heart failure in specifically Black patients) (Roberts 2011). And regularly, people of color are wrongly diagnosed with conditions solely based on presumptions about how their race and ethnicity predisposes them to ill-health (Shim 2005; Aronowitz 2008).

In a similar vein, there is a long history of gender making its way into medical pathology. As early as the 18th CE, ideas around sex, gender, and sexuality constituted medical pathology, setting apart the 'normal' from the 'deviant' (Foucault 1976: 43). Individuals were considered 'normal' only if they were heterosexual men and women. Children were seen as sexual creatures that needed medicalized control, and 'perverse pleasures' were pathologized as abnormal behaviors in need of treatment. Women in particular were brought under medical scrutiny based on the assumption of their hysterical female bodies (Foucault 1976: 93-95).

These ideas about gender being embodied, and accordingly characterizing normal versus deviant existences, persist in medicine. For instance, as trans individuals navigate potential gender-affirming procedures, medical institutions, surgeons, and insurance companies hold significant influence over deeming what counts as a necessary and appropriate procedure for a particular individual given their gender identity, genetics, and neurological presentation (Gonsalves 2020; Meyerowitz 2004; Gill-Peterson 2014). Scientists regularly interpret sex chromosomes, sex cells (i.e., eggs, sperm), and gonads (e.g., testes, ovaries) as dichotomous (male/female) and as having

traditionally gendered functions and characteristics, regardless of mounting scientific evidence suggesting more variability in physiological sex (Ainsworth 2018) and the social rather than biological basis of gender (Karkazis 2012). Stereotypes of women's passivity compared to men's strong-willed deliberation are imbued in scientific literature that explains eggs as receptacles that "depend on sperm for rescue," when indeed research shows fertilization to be a far more cooperative process (Martin 1991: 495-499). Similarly, the X and Y chromosome are assumed to have biological underpinnings of female- and maleness, respectively. Each chromosome is ascribed opposing gendered traits in scientific literature, with the X supposedly leading one to be "sociable, controlling, […], monotonous, and motherly" while the Y is "macho, active, clever, wily, dominant […] (Richardson 2012: 911-912)." Classifying sex, gender, and sexuality as physiological identities, and distinguishing which presentations are 'normal,' has long been, and continues to be, a preoccupation of medicine.

The Life of Joan/John 1965-2004

The way medical institutions determine a biologized relationship between sex and gender can have severe consequences. While there are many examples that illustrate such harms, I will rely on the life of Joan/John (pseudonym) (Butler 2001, Fausto-Sterling 2013). John was born in 1965 in Winnipeg, Canada as a phenotypical male, with male presenting genitalia. When he was merely 8-months old, his penis was irreparably burned and severed during a surgical operation similar to a circumcision. Struggling to accept this outcome for their child, his parents turned to prominent psychologist John Money after watching a television program where Money discussed the supposed benefits of transexual and intersexual surgery (i.e., sex reassignment surgeries and hormonal treatments). John's parents met with Money at Johns Hopkins University, where Money strongly recommended that the parents proceed with a surgery to feminize infant John's genitals and raise the child as a girl. Following this, John's testicles were removed, what remained of the penis was to be considered a clitoris, and plans were made to build a vagina once the child was older. Now Joan, the child was to live as a girl, and would attend regular 'counseling' sessions with Money where they would be taught how to act as a girl should. (Butler 2001)

At the time, Money was able to enlist support from well-known feminists for his approach to Joan/John's situation, based on his skewed presentation that gender could be engrained via nurture rather than determined by nature. Feminists including Suzanne Kessler and Wendy McKenna turned to Money's treatment of Joan/John to exemplify the idea that one's anatomy need not be their destiny (Kessler and McKenna 1985). However, others, including Judith Butler (2001) and Anne Fausto-Sterling (2013), sharply critiqued Money's approach to Joan/John, fundamentally resisting that a person needed to be forcefully conformed to a binary and biologized formulation of sex and gender at all. In explicating Joan/John's life, and larger ideas around variable sexed features and their distinction from lived gender, I rely on Butler and Fausto-Sterling's perspective.

For Money and the medical professionals involved in John/Joan's care, their concern was less about the child's self-perceived gender identity and more about societal perceptions – the appropriate identity the child *should* have given the botched surgery that ruined their penis. They believed, and imposed on the family, that gender should be taught in alignment with how the genitals presented for a 'normal' life experience. These professionals were preoccupied with defining normality and weaponized societal shame in order to insist parents do the 'right' thing for their child (Butler 2001: 626). These attitudes prevailed beyond John/Joan's experience, as medical

literature from the time holds that a boy must have a penis big enough for him to pee standing up, so that he can "feel normal when they play in little boys' peeing contests." Relevant to John/Joan's case, when a penis was 'too small' surgeons tended to follow a rule-of-thumb saying, "it's easier to dig a hole than build a pole," referencing their preference to reassign these individuals to female genitalia and insist they be raised as girls (Fausto-Sterling 2013: 222). The medical obsession with normality being an alignment between sexed genitalia and psychosocially experienced gender reflected not a genuine concern for each patient, but rather for upholding scientists' and doctors' biases around traditional societal norms (Fausto-Sterling 2013).

As Joan approached their teenage years, they started to realize that they did not "feel like a girl" and identified more with how the boys around them were allowed to live. Joan's discontent with being forced to live as a girl grew. They refused to take estrogen, hated developing breasts, and even ran screaming from a room where Money forced them to look at graphic images of vaginas. Eventually, a group of psychiatrists and doctors took notice of Joan's misery. They offered Joan gender-affirming procedures to, once again, appear male, reversing the decade of treatments Money had insisted they endure. Now John, he willingly took male hormone shots, had breasts removed, and had his penis reconstructed. John lived this way for several decades, but consistently grappled with severe depression as a result of his childhood trauma. At the age of 38, John died by suicide. All the while, Money profited immensely. He publicly claimed that Joan/John's sex and gender reassignment had been an unparalleled success showcasing how sex could be 'constructed' and gender taught accordingly, continually recommending this forceful medicalized approach for other intersex children. (Butler 2001)

The life of John/Joan brings into stark relief the consequences of a narrow and essentialized view of gender's relationship to the sexed body. It makes imperative an understanding of how experts wield medical tools and scientific claims toward biased sociocultural perspectives. Importantly, it clarifies that ideas around sex chromosomes, genitalia, gonads, and their relationship to one's gender identity are not objective truths, but rather constantly shifting perspectives that arise in part from scientists and doctors who imbue their own prejudices in the production of medical knowledge (Martin 1991; Richardson 2012). Fausto-Sterling (2013) terms these experts "medical managers," reflecting the power and authority they cast when determining the sexed and gendered lives of patients. Joan/John's life reminds us of the need to question and resist conventional notions of what is medically classified as 'normal' or 'abnormal' around sex and gender – and of the possible tragic consequences of failing to do so – especially with regard to reducing one's identity and life experiences to their physiological characteristics.

Bringing Sex Chromosomes to the Forefront with Genetic Testing

Fausto-Sterling (1993, 2013) is among scholars who have outspokenly challenged imposed surgical interventions that conform intersex infants' genitalia to a binary vision of sex, showing through her work the existence of at least five sexes.[33] Today, these critiques can also be extended beyond medicine's concern over ambiguous genitalia, as genetic technologies like Non-Invasive Prenatal Testing (NIPT) give rise to new 'problems' and new opportunities for medical managers to continually shape medicalized views on sex and gender. A screening tool, NIPT identifies

[33] Aligned with Fausto-Sterling's research, there is robust scientific evidence supporting the idea that embodied sexed features do not solely represent a male/female dichotomy, that there is much greater variability in how sex presents within one's physiological features (Ainsworth 2018).

various chromosomes; it is routinely and effectively used during pregnancy to screen fetuses for chromosomal conditions including Down (trisomy 21), Edward (trisomy 18), and Patau Syndrome (trisomy 13). NIPT can also identify a fetus' sex chromosomes, which has become an increasingly pertinent reason for its use. In 2022, over 3 million NIPT tests were performed worldwide – a two-fold increase from 1.5 million tests just two years prior in 2020 – with 600,000 tests occurring in the US alone (the country with the highest NIPT use) ("Genomics" 2022, "Non Invasive" 2022). Given the growing reliance on NIPT, and its ability to identify fetal sex chromosomes, how do prenatal medical managers interpret fetal sex chromosomes within the constellation of sexed biophysical traits (e.g., genitalia, gonads) and the resulting relationship to notions of gender?

As NIPT reveals sex chromosomes, sex chromosome aneuploidies (SCAs), a set of previously underdiagnosed conditions, are now an increasingly visible diagnostic category at the prenatal stage (Navon 2019). Essentially, patients diagnosed with SCAs present with sex chromosomes that don't conform to the typical XX (female) or XY (male), once more medically delineating the 'normal' from 'abnormal.' Sex chromosome variations include Klinefelter syndrome (47,XXY), Jacobs syndrome (47,XYY), trisomy X (47,XXX), and Turner syndrome (45,X), among several others. Importantly, SCAs range in their severity and often present as mild, if noticeable, conditions, in those who are born with these variations. Current research estimates that SCAs are among the most common genetic variations, affecting 1 in 500 individuals. Because sex chromosome variations do not often result in differing appearances, and symptoms can be rather mild, only 25-30% of individuals are ever diagnosed (AXYS n.d., Samango-Sprouse 2016). As such, providers and patients are not always certain what to make of these diagnoses.

Still, increasing illumination of these diagnoses via NIPT inevitably raises questions for expecting patients and providers around a to-be child's well-being as it relates to sex and gender. A growing aspect of prenatal care and reproductive decision-making, SCAs reinvigorate essentialized views about gender as an identity determined by one's biophysical traits (i.e., chromosomes), potentially positioning those with atypical sex chromosomes as societal deviants (Foucault 1976). In what follows, this chapter critically examines the re-emergence of the SCA diagnostic category as a feature of routine NIPT use, questioning the notion that the body is 'truth' when it comes to gender (Fausto-Sterling 2013; Butler 2001) and exploring the consequences of pathologizing a sex and gender relationship.

A Brief History of Sex Chromosome Aneuploidies and their Diagnostic Implications

From its inception, genetic testing has informed a medicalized understanding of sex and gender, often leading to ideas that X versus Y chromosomes render fundamental gendered differences in individuals (Richardson 2012). Genetic technologies started making SCAs visible as 'abnormalities' around the 1950s and 1960s. As a result, the SCA diagnostic category has captured a medical imagination which involves pathologizing sex and gender to make claims about an individual's health, well-being, and supposedly biological predispositions.

Among the first conditions to be characterized were Turner (45,X) and Klinefelter (47,XXY) syndromes in the 1950s, each suggesting that the X chromosome had an atypical presence and would impact an individual's femininity. Following these findings, in 1959, trisomy X (47,XXX) was identified and referred to as "Super Female" syndrome, due to the presence of an extra X chromosome. Soon after, Jacobs syndrome (47,XYY) was detected in 1961 and discussed as the

"Super Male" syndrome that caused aggressiveness, with significant consequences for how these boys and men were perceived in society (Navon 2019: 69). Below, I explore the history of Jacobs syndrome to highlight how a sex chromosome diagnosis can shape one's life experiences, despite these conditions not necessarily *causing* those affected to behave in anti-social or harmful ways. Individuals' can be negatively impacted simply because findings of their sex chromosomes make them visible as 'gender deviants' and lead their behaviors to be pathologized accordingly, without consideration for the structural pathways that consequentially shape their experiences.

Early studies of Jacobs syndrome established a stereotype that those with this condition were aggressive, anti-social criminals. In 1965, researcher Patricia Jacobs began studying prison and asylum populations in Scotland. With a landmark publication in the renown scientific journal *Nature* – "Aggressive Behavior, Mental Sub-normality and the XYY Male" – Jacobs and her colleagues declared that 47,XYY was linked with criminal behaviors, thus beginning the enduring legacy of this genetic variation being considered a 'criminal chromosome' (Navon 2019: 69). Jacobs' research teams went on to study "mentally subnormal male patients with dangerous, violent or criminal propensities" in Edinburg, aiming to establish a connection between 47,XYY and criminality. Where today their sampling strategies have been criticized as selective and biased, at the time their finding that 12 of 197 of these 'dangerous' men had Jacobs syndrome was heralded as a major outcome (even though, outside the penal setting, it was thought that only 1 of 2000 males presented with 47,XYY at the time). By 1966, articles in leading journals like the *Lancet* lauded these findings, solidifying the notion that XYY males have in-born criminal tendencies and are thus fated for mental-penal institutions. This paved the way for decades of research endeavoring to characterize the XYY 'Super Male.' Men with Jacobs syndrome were identified as being especially tall and their so-called criminal tendencies were considered a geneticized psychiatric disorder (Navon 2019: 70-71). Over time, these men were labelled as mental defectives, delinquents, aggressive, and violent – tropes that persist today when expecting parents and providers discuss 47,XYY.

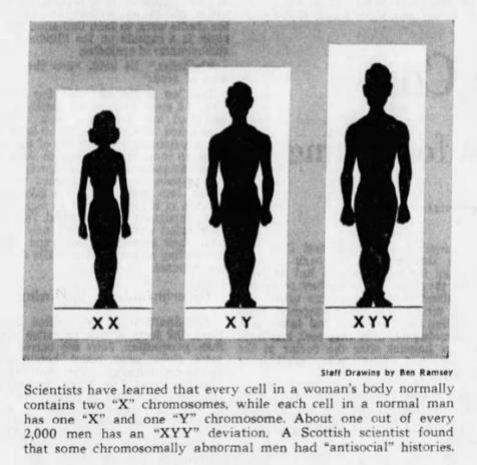
One way these misconceptions around Jacobs syndrome became visbile was via institutionalization in the legal system. In the late 1960s and 1970s, the idea of a 'criminal chromosome' started to appear in courtrooms around the world, in an effort to defend men who had committed violent crimes. To garner lenient sentences, attorneys would establish that defendants were, to some extent, incapable of sound judgement and naturally prone to dangerous aggression due to their Jacobs syndrome (Chandler and Rose 1973, Fox 1971). One of the earliest instances of this reasoning came in 1968, in a French court trying Daniel Hugon for the brutal strangulation and murder of a 62-year-old sex worker. At the trial, French geneticist Jerome Lejeune (who uncovered the association between trisomy 21 and Down syndrome in 1961) testified that Hugon "had been doomed to be a sick man with a hereditary inability to exercise normal responsibility." The jury gave Hugon a much lower sentence of 7 years due to his 'criminal chromosome' (Navon 2019: 71-72).[34] In the United States, there are at least two horrific rape and murder cases – involving defendants Sean Farley in 1968 and Kevin Goode in 1969, both in Queens, New York – where defense teams mobilized the criminal chromosome argument

[34] In the same year as the Hugon case, an Australian court acquitted Laurence E. Hannell of stabbing and killing a 77-year-old widow on grounds of insanity, owing to his 47,XYY variation. The 'criminal chromosome' line of reasoning, as supported by Patricia Jacobs' research on men in mental-penal institutions, became an established precedent for legal teams defending men who had committed violent acts in countries across the world (Fox 1971).

(although, to no avail). In all these cases, the men were described as tall, anti-social, aggressive, and lacking in mental capabilities. Their genetic variation was framed as determining their horrifying actions. (Baker 1969; Saxe 1969)

Even though these arguments were not always successful in the courtroom, they managed to firmly ground a causal, geneticized connection between Jacobs syndrome and violent behaviors in the public's imaginary (Navon 2019: 73). Each time the criminal chromosome line of reasoning was harnessed, it fueled an essentialized understanding of what it means to be 'Super Male,' pinpointing the extra Y chromosome as driving serial killers, rapists, and murderers. Indeed, popular media at the time, including The New York Times, fervently covered the 'XYY criminal chromosome.' Newspapers ran headlines like "Genetic Abnormality Linked to Crime" alongside articles discussing the genetic penchant 'Super Males' have for violent and dangerous behaviors, essentially distinguishing the gender 'normal' from 'abnormal' in the public's mind. ("Genetic" 1968; "Man" 1971)

A 1968 illustration in The Courier-Journal (Louisville, Kentucky) depicting XYY as a deviation from 'normal' sex chromosome presentations:



Noting the uptick in media attention given to 47,XYY as well as its recurrent appearances in legal defenses, Patricia Jacobs penned a letter in 1968 expressing her uncertainty around the conclusions about Super Maleness. She admitted to a sampling bias in her initial study and underscored the lack of reliable estimates of 47,XYY in the general population, which precluded an effective comparison between affected men who did and did not have criminal histories. Jacobs also revealed that there was no single, deterministic presentation of 47,XYY; one's symptoms and behavioral presentations could range immensely, perhaps owing to social experiences and structural effects. In other words, one could not definitively say that a man with Jacobs syndrome would be aggressive, violent, or anti-social. As Jacobs shed doubt on her original conclusions, she called for more studies around the developmental effects of sex chromosome variations – a sentiment echoed throughout the 1970s as researchers gradually uncovered that 47,XYY was far more common that initially estimated, affecting around 1 in 300 births. (Navon 2019: 73-75)

Regardless, the 'criminal chromosome' myth had taken hold both on scientific and public fronts. Although research had established that only a minority of crimes were committed by those with

47,XYY, and scientists were questioning the notion of a criminal chromosome, there continued to be a preoccupation with unraveling a genetic explanation for unwanted behaviors and aberrant maleness. Reflecting on discussions at the 1968 American Society of Human Genetics meeting, Noble Prize-winning biologist Joshua Lederberg insisted on continuing to study the genetic predispositions of those with Jacobs syndrome. In a Washington Post article, he wrote, "we have no idea about the roots of the problem or just what fraction of XYYs will make such miserable failures of their own lives and inflict so much harm on others." Stating that the scientific community needed to adequately understand "the biology of violence," Lederberg ignited studies of the criminal chromosome for decades to come. These included a 1973 National Institutes of Health sponsored review of 47,XYY, which referred to the chromosomal variation as the 'Hyper-Male' complex, linking "an extra Y chromosome" with "heightened masculinity, […], and powerful aggressive tendencies" (Navon 2019: 77). Accordingly, scientific experts propelled the notion of a dangerous gender deviant as someone with 'too many' Y chromosomes and naturalized gendered differentiations based on sex chromosomes. These misinformed research agendas further marginalized a growing group of affected men, with lingering effects on narratives essentializing one's behaviors to chromosomes.[35] (So 2022. Navon 2019: 75-76)

While most research about the 'criminal chromosome' had been phased out by the late 1970s, its grounding in the scientific literature continues to provide traction for medical myths and studies about SCAs (Navon 2019: 83). Today, similar assumptions about SCAs show up as early as the prenatal stage, as the routinization of NIPT has made learning about sex chromosome variations more common. Rather than criminalizing those with particular variations, the contemporary objective for testing fetuses for SCAs is largely geared toward early diagnosis and childhood interventions for possible behavioral and developmental needs – an agenda supported by the advocacy group Association for X and Y Chromosome Variations (AXYS) (Navon 2019: 89, 92). However, these diagnoses are also engendering discussions about what it means to present one's gender appropriately or 'normally,' creating instances where those with sex chromosome variations are pathologized as potentially not fulfilling societal expectations of how sex chromosomes inform gender identities. SCA findings on NIPT results often lead to complex and ambiguous situations wherein expecting parents and providers range from being unclear on the diagnostic implications to more directly pathologizing an affected child's gendered life experience. In some instances, expecting parents do terminate for SCAs, particularly Jacobs syndrome given its misconstrued history, even though these conditions are increasingly common and often do not noticeably compromise a child's life (Navon 2019: 85, 90). This renewed interest in SCAs, in large part owing to reproductive medicine's growing reliance on NIPT, has recast a diagnosis that otherwise may never have been consequential into a "genetic disorder" centered on normative conceptions of sex and gender, with a "mandate for early intervention" (Navon 2019: 213).

NIPT as 'the Gender Test'

[35] In the United States, these studies had disproportional racialized effects for young men of color. Prominent research institutions including Harvard Medical School, Boston Children's Hospital, and John's Hopkins University partnered with federal agencies to study tens of thousands of young Black boys for 47,XYY. Black boys in juvenile jails were genetically tested and studied without their families' consent, and their results given to juvenile correctional agencies as evidence for the boys' criminal propensities, intentionally jeopardizing the children's futures (Jackson et. al 2023; Navon 2019: 79-81).

The technological capacities of NIPT mark a new milestone in medicine's preoccupation with embodied sex and gender – and, accordingly, the delineation of "normal" and "aberrant" existences based on sex chromosome variations and 'discordant' genitalia. A prenatal genetic technology, NIPT uses a maternal blood draw to screen fetuses for chromosomal variations, providing results as the 'percent chance' that a fetus has a particular condition. Unlike prior prenatal screenings, it also detects sex chromosomes, and thereby identifies the possible presence of sex chromosome aneuploidies – a piece of information that pregnant patients did not routinely have access to before NIPT's introduction.[36]

While physicians may value knowledge about fetal sex chromosomes to understand whether a pregnancy presents with an SCA necessitating heightened prenatal care, for many pregnant patients, knowing their fetus' sex chromosomes often equates to learning the 'gender' of their to-be child. This is largely because commercial genomics companies vastly marketed NIPT to patients as the test that can 'tell you whether you're having a boy or a girl as early as 9 weeks' in one's pregnancy ("Early" 2023, Bradley 2021). Thus, the 'gender test' terminology allows non-genetics providers to easily relate to patients, as this NIPT descriptor is well-recognized among pregnant patients. Moreover, it encourages many patients who want to know their to-be child's 'gender' (i.e., sex chromosomes) to pursue testing, which in turn benefits physicians who can use the robust genetic data to manage these pregnancies. NIPT has become so familiar as the 'gender test' that many pregnant patients eagerly opt into testing to organize 'gender reveal' parties (Richards 2019, Langmuir 2020). However, while providers may be getting clinically useful genetic data, repeatedly framing NIPT as the 'gender test' perpetuates inaccurate notions about its purpose.

As NIPT becomes popularized as the 'gender test' it opens the door for more routine use of the technology to identify sex chromosomes, yet without patients and providers having a thorough understanding of possible SCA results and their clinical implications. This leads to misinformed assumptions about those with SCAs as 'gender deviants,' anxieties around whether or how a child's health will be affected, terminations of wanted pregnancies that may indeed be healthy, and increased investments into broadening NIPT's SCA capacities. Taken together, these outcomes continually reinforce SCAs as a medically concerning diagnostic category, despite scientific evidence and health advocates insisting that most SCAs do not significantly compromise one's health, and narrow societal tolerance of those with variable sexed and gendered existences (AXYS n.d., Samango-Sprouse 2016). In what follows, I illustrate how NIPT becomes skewed into the 'gender test' and the consequences this limited understanding of sex chromosomes as conflated with gender expectations has on pregnant individuals and their providers.

Ideally, a patient should consult a genetic counselor prior to having their blood drawn for NIPT. Almost all providers interviewed agreed that this would be the best approach, even though some considered it unfeasible due to a lack of resources and time constraints. During this pre-test consultation, genetic counselors explain NIPT to expecting parents, discussing how the test works,

[36] NIPT is a screening test, and not diagnostic test. As such, its results (which are presented as the likelihood, or percent chance, that a fetus has a particular condition) need to be confirmed via diagnostic testing like amniocentesis or chorionic villus sampling (CVS), both of which involve invasive methods to collect necessary samples.

the conditions it screens for, and its efficacy for identifying each condition. Crucially, the pre-test consultation allows space for patients to ask questions and make an informed decision regarding prenatal testing. When this process works well, as it does in the genetics case I observed below, the consultation enables patients to make informed decisions.

Genetic counselor Raquel meets with expecting parents Robert and Linda, who are 4 months into their pregnancy. The pregnancy is long-awaited and much desired, as the couple underwent several rounds of in vitro fertilization before becoming pregnant and express how excited they are to have come this far.[37] They have been referred to genetic counseling by their Obstetrician Gynecologist because Linda is over 35 years old, and therefore considered to have a high-risk pregnancy.[38] The Obstetrician Gynecologist introduced prenatal genetic testing to the couple but did not go into much detail aside from providing a handout and directing them to their genetic counseling session.

At the start of the genetics consultation, Linda makes her position on testing the pregnancy clear: "In general, I think with testing we err on the side of not wanting to know because it can cause more anxiety, and I know false positives happen, and we just don't want to deal with all that." Raquel is supportive but also wants to make sure the couple has sufficient information about the contemporary prenatal testing setting before they make a final decision. She tells them that "testing has come a long way in the past years, so false positives have gotten a lot better" and distinguishes the first and second trimester screens of the California Screening Program, which has a high false positive rate, from NIPT (Genetic Counseling Ethnography 2021). Robert and Linda lean into the conversation about NIPT, so Raquel continues:

> "NIPT looks for little pieces of chromosomes in your blood that come from the placenta, typically that's the same as the chromosomes in the baby. So, if we see a lot of pieces of chromosome 21, then that could mean Down's Syndrome. If we see a normal amount of chromosome 21, that means that baby is probably doesn't have Down's Syndrome. NIPT looks for the 5 main chromosomal conditions, including Down's Syndrome, trisomy 18, 13, *and sex chromosomal aneuploidies*. For trisomy 18 and 13, these are birth defects that a lot of children unfortunately don't usually survive. *Sex chromosome aneuploidies are similarly genetic conditions that are birth defects, but those are much milder. NIPT is not 100% accurate, no blood test is. But it detects 93-99% of babies with those conditions.* It can have false positives, but not as much (as other prenatal screening tests) (Genetic Counseling Ethnography 2021, *emphasis added*)."

Raquel presents NIPT as a medical tool. Specifically, she describes how the screening detects sex chromosome aneuploidies, as opposed to any mention of sex or gender. Robert and Linda remain very engaged and ask numerous questions. Robert inquires, "In terms of preparing mentally, we are 5 months out from delivery and there's already a lot to do. Do 5 months make a big difference

[37] For unclear reasons, Robert and Linda are considering testing options at a much later timepoint than most expecting parents in California. NIPT, in particular, is offered to patients at 9-13 weeks of pregnancy.

[38] Referring high-risk patients to genetics for an overview of testing is a standard prenatal practice, as certain parental factors including maternal age can increase the likelihood for fetal chromosomal variations and potential delivery complications.

in knowing or not knowing this information? How much more can we get prepared now than when baby is born?" (Genetic Counseling Ethnography 2021). Raquel provides a thorough response:

> "That's a good question. It does depend on the condition. So, for sex chromosome abnormalities, those are milder, you might not even notice, so not much preparation (is required). But for something like Down's Syndrome, you can be preparing. Like if there's heart defects that come with Down's Syndrome, which sometimes happens, we can know to do more scans or attend to (the baby) when they are born. Or, if you find something more severe, like trisomy 18 or 13, where we know they might not survive long, we can make arrangements for palliative care, or a c-section or a vaginal birth. We can just have a plan, so we are prepared. For something like Down's Syndrome, it might also affect the delivery method. Really, all this testing could be helpful for you both to be emotionally prepared, do your research about these conditions, but also for doctors to be prepped in the delivery room for how best to support your baby. (Genetic Counseling Ethnography 2021)."

Perhaps the most useful aspect of pre-test counseling, Raquel provides the expecting parents with a gamut of ways the NIPT results could be useful, both for parents' and providers' preparation.[39] Robert and Linda continue to ask questions about NIPT and other testing options, including amniocentesis and newborn screening, weighing these against their reproductive priorities. Ultimately, Robert turns to Linda for a decision, and she quickly answers, "We won't do the amniocentesis. For now we're still probably going to wait until delivery for any testing" (Genetic Counseling Ethnography 2021). Raquel respects their choice and continues to hold space for questions, all of which she responds to by centralizing the couple's reproductive values. Nowhere does she frame NIPT as the 'gender test,' but instead describes it as a medical tool.

Patients can expect the supportive and informative pre-test counseling Robert and Linda received from Raquel *if* they are routed to a genetic counselor or other genetics providers (e.g., geneticist, maternal fetal medicine specialist). Unfortunately, this is not the reality for most pregnant patients, as evidenced in data obtained from interviews with patients and providers as well as patient discussions on Reddit forums (a popular website where users can engage in specific discussion threads).[40] In these data, we see that most patients are not interacting with genetic counselors before testing, and instead receive information about prenatal genetic testing from non-genetics providers – Obstetrician Gynecologists or nurses – who often refer to NIPT as the 'gender test.' In fact, almost none of the patients I interviewed saw a genetic counselor *before* their blood had been drawn for NIPT, and many did not see a genetic counselor at all. In many practices, physicians deem it unfeasible to refer each patient to genetic counseling. Some discuss this as a time constraint issue: because they see so many patients, they need to streamline prenatal care unless a genetic test

[39] Notably, Raquel does not discuss the options of termination or further testing with Robert and Lina, which is a common discussion on other pre-test consultations. She tells me this is because the couple established their hesitation toward prenatal testing and were not considering termination given the fertility challenges that they waded through. Their pregnancy was also nearing California's cut-off for electing termination.

[40] Reddit is a popular website with threads or 'subreddits' geared toward specific discussion themes. Users can be a part of discussion communities focused on their needs and interests, including specific threads/subreddits for patients to discuss prenatal genetic testing. Users can post their experiences or questions, respond to others via comments, as well as react using features like 'up-votes' (similar to 'likes') ("Reddit" n.d.)

result indicates cause for concern. Others state that there are not enough resources to refer each patient; not every reproductive practice is fortunate to have an in-house or accessible genetics clinic. And, while most non-genetics specialized physicians understood the value of pre-test genetic counseling, many also saw NIPT as being 'simple enough' that they could explain it to patients themselves. As a result, patients tend to receive overly simplified, and at times incorrect, information, about prenatal testing from non-genetics providers.

In almost all cases, when NIPT was presented to pregnant patients by ObGyns or nurses, it was discussed as 'the gender test,' even though these providers understood that NIPT had little if anything to do with gender. Still, these providers are motivated to have patients pursue NIPT because of its efficacy in identifying complex chromosomal conditions that may require additional medical attention. As such, non-genetics providers often present NIPT as the 'gender test' because this terminology resonates with patients, who they assume will want to know the 'gender' of their to-be child. If patients opt-in to learning their to-be child's 'gender,' medical providers can access the larger genetic data to better manage a pregnancy. In this way, NIPT becomes reinforced popularly as the 'gender test,' even while non-genetics providers understand that this is not its purpose. Below, patients describe how they were introduced to NIPT by non-genetics providers:

> "(Our Ob) said that we could know the genetics and the *gender of the baby* sooner, and then she said that it would look for like Trisomy 18 Trisomy 21, those sorts of things. And then she sent us home with a pamphlet to read. And in the reading of the pamphlet is where I've found that it is not a black and white, your child will have this no they won't, (but) it's a percentage, and because of the percentage is why I elected to not to do it (Cheryl Interview 2021, *emphasis added*)."

> "(The nurse) mentioned that with the NIPT *you could find out the sex of the baby quicker*. Like at around 2 months. Usually you find that around 4 months. *So that was one thing they did try to emphasize. Later on, one of the receptionists tried to upsell me on the NIPT* when I was coming in for one of my appointments. She asked if we were going do the NIPT. I told her, 'No, I can't because my insurance doesn't cover it.' And then she's like, *'Oh, but you find out the sex of the baby.' I was like, 'Yeah, that's enticing but I'll just wait another 2 months'* (Nalini Interview 2021, *emphasis added*)."

Most striking is the difference in how Raquel comprehensively discussed NIPT as foremost a medical technology compared to how Cheryl and Nalini learned about the screening as a way to know 'gender' from their non-genetics providers. For both Cheryl and Nalini, NIPT was presented as a test that would tell them their child's 'gender' (fetal sex) quicker than other prenatal tests. In Nalini's case, clinic staff also tried to emphasize gender to encourage her to elect NIPT even though she declined the screening, illustrating how providers may try to garner genetic information about chromosomal conditions through NIPT by tapping into expecting parents' desire to know their fetus' 'gender.'[41] In other interviews, patients recalled or distinguished NIPT from the slew of other prenatal tests because it was 'that test that checked for gender': "I remember they did a lot of testing. For one, they tested for 3 trisomy conditions and tested to see what the gender or the sex of the baby is (Bethany Interview 2021)." While Cheryl and Nalini declined NIPT, providers

[41] Note, Nalini did her early pregnancy consultations outside California, in Texas, where insurance coverage for NIPT is not yet as routine.

described that most patients pursue the screening – not least because it will reveal their fetus' sex chromosomes (or, as patients describe, their child's gender).

As 'the gender test,' NIPT makes discussions around sex and gender that much more visible during pregnancy, an experience that is already marked by revelations about the child being a boy or girl. Moreover, when non-genetics providers increasingly present NIPT as the 'gender test,' it becomes framed as less of a medical tool that can identify chromosomes, including sex chromosomes, for possible genetic conditions. Instead, the test becomes a way to emotionally appeal to expecting parents desiring information about their to-be child's 'gender.' We see this emotionality surrounding NIPT in the Reddit post below, where a patient discusses her excitement around learning her child's 'gender.' She emphasizes how her pregnancy now 'feels real,' highlighting the powerful social effect that sex and gender have on grounding our relationships and expectations of one another (in this case, between parents and their child to-be):

> "We had the Harmony NIPT done, and honestly, this whole thing has been feeling so unreal […] that I had zero anxiety about the results, because how can I be anxious about something that doesn't feel real? *The main reason I wanted the test was to find out the gender, because I felt like once we know that, then it'll feel real. This isn't just an abstraction anymore, it's our child, and we've made it all the way this far.* So I was completely blindsided when I missed the call from the GC and went to listen to her voicemail, and the first thing I heard was, "I have the good news you've been waiting for..." […]. *I didn't realize I was holding my breath about this. I haven't found out the gender yet.* I asked her to write it down in a card for [my significant other] and me, and I'm going to pick it up after work, and we're going to go out to dinner tonight and open it together. *But... while I'm excited to find out if we're having a little boy or a little girl... we've cleared another hurdle, y'all. We're past another mile marker. All the tests came out low-risk, and we're on our way to having a healthy kiddo* […]" (Reddit Patient Forum 2021, *emphasis added*)."

This patient illuminates how NIPT has become deeply situated as the 'gender test,' rather than a medical tool screening chromosomal conditions, in expecting parents' minds. When patients opt into NIPT it is often *because* they want to learn about their fetus' sex chromosomes; the results around chromosomal conditions, while worthwhile, seem to be a secondary consideration. For example, when asked why she and her husband went ahead with NIPT during an interview, Julia told me: "We definitely wanted to know sex. We were impassioned about that, but I also liked the idea of screening for a certain set of particularly common conditions, because I felt they would have given us the information to have a conversation as a couple about how to proceed in light of that information in time (Julia Interview 2021)." Similar to the Reddit poster above, Julia describes desperately wanting to learn about fetal sex, with knowledge about the fetus' genetic health being an additional benefit rather than the primary motivation for pursuing NIPT.

NIPT's popularity as 'the gender test' can be frustrating for genetics providers, especially genetic counselors and maternal fetal medicine physicians, whose work centers around understanding how genetic markers impact prenatal care. These genetics providers described that genomics companies and non-genetics providers promoting NIPT as a 'gender test' obfuscates that the test is actually an integral medical technology aiding prenatal care. This not only compromises informed consent, but also leads to situations where patients receive results about chromosomal conditions that they

were unknowingly being tested for – particularly, SCAs. Further, treating NIPT as a 'gender test' downplays genetic counselors' roles as genetics providers, as patients come to see them as primarily responsible for revealing the to-be child's sex. Amy, a genetic counselor with over 10 years of experience, describes her exasperation with NIPT's rapid uptake as 'the gender test':

> "People will literally call genetics and say, 'I want to schedule an appointment for the gender test.' It drives us crazy because it's not a gender test! It's a medical test that gives you genetic information about the fetal chromosomes, and gender is not determined by sex chromosomes alone. And I'm also like, 'don't yell at me that your results are late, and your gender reveal party has to be on whatever day. Because I don't care. I am not a party planner!' One lady called me and they're like 'Can you call the bakery with my test results?' and I'm like, 'No! I am definitely not doing that!' Or 'Can you call my cousin and tell her the sex of the baby?' We literally had to come up with a policy in our office about who we will tell the sex of the baby to because we get so many of these ridiculous requests (Amy Interview 2021)."

Framing NIPT as 'the gender test' misrepresents the screening's intended medical purpose and lead to incomplete patient education. Many of these issues emerge when patients are not appropriately counseled by genetics providers prior to testing. Specifically, genetics providers repeatedly pointed to issues with how patients understood the potential for SCA findings. While patients associated NIPT with knowing their child's 'gender' via sex chromosomes, they did not always realize that the test could therefore also identify SCAs or provide inconclusive SCA results. Genetic counselors described these challenges as especially salient for those who did not see genetics providers prior to testing:

> "I had a patient that was high risk for either XXY or XYY. But the patient did not want to know the predicted sex of the baby and the (Obstetrician Gynecologist) had not taken that opportunity to specify that if we do (NIPT), there is a possibility that we will learn the predicted sex of the baby in the context of a medical diagnosis. […] And so they came to us being high risk for this condition, that they didn't know they were even being tested for, and that they didn't want to know about. They wanted to continue the pregnancy. But now the experience has been kind of poisoned by medicalizing this information that, most likely, is not going to be a huge impact on that child's life (Leena Interview 2021)."

> There is a lot of misunderstanding. Most individuals think (NIPT is) diagnostic and it's not. I don't think it gets conveyed (by their Obstetrician Gynecologists) that NIPT is a screening test […]. Sometimes we'll see people who are *positive NIPT for maybe not Down Syndrome, but for a sex chromosome aneuploidy. And that doesn't always get translated to the patients, that that's also being tested for*. […] And patients will say, 'I wasn't told they were testing me for this as well' (Rosemary Interview 2021, *emphasis added*).

Because non-genetics providers rely on the relatable 'gender test' terminology when discussing NIPT with patients, many of whom do not receive thorough genetic counseling prior to testing, patients do not always realize that the test can return a possible SCA diagnosis. Without adequate preparation, a patient's pregnancy experience can be 'poisoned,' as Leena describes, by this information, which may go onto have consequences for one's reproductive decisions. As such, the

rapid routinization of NIPT as the 'gender test,' rather than foremost a medical tool, leaves much to be desired when it comes to informed and equitable prenatal screening. Not only does this approach to screening bring sex and gender to the forefront of pregnancy, inaccurately essentializing gender to one's sex chromosomes, but it also creates a new 'problem' – the prenatal diagnostic category of sex chromosome aneuploidies and the related implications of gender expectations being translated into medically relevant concerns.

Sex Chromosome Aneuploidies as a 'New Problem' in Prenatal Care

Using prenatal technologies to define a fetus' sex is not a novel medical practice. As early as 1958, physicians have used ultrasound machines to image fetuses in-utero, which included visualizing their developing anatomical features (i.e., genitals, gonads) to classify their sex per the male/female binary. Sex chromosomes, too, have been identifiable (in insects) since the early 1900s, while genetic technologies to do so in human reproduction became commonplace in clinics during the 1970-1980s with amniocentesis and CVS (Campbell 2013, Abbott et al. 2017). These technologies long predate NIPT's 2011 release into the U.S. consumer health market. What the NIPT era has engendered, however, is the *widespread* use of a genetic technology across pregnancies to *routinely* identify fetal sex chromosomes – hence its popularity as 'the gender test.' As a result, the technology also detects SCAs at a much higher frequency than ever before, unveiling these diagnoses as a problem that providers and patients now need to contend with during the prenatal stage.

Prior to NIPT, a doctor would have relied on ultrasound to classify fetal sex and 'normal' development of male or female anatomy. They would only resort to genetically testing for sex chromosomes in the case that atypical imaging prompted further investigation. However, with today's popular use of NIPT as a 'gender test,' there is more opportunity for an SCA diagnosis to emerge prenatally, as the screening can place sex chromosome findings in conflict with a physician's classification of fetal sex per ultrasound. Dr. Harris describes this tension, highlighting the seeming novelty of SCAs as they coincide with the widespread implementation of NIPT:

> "As NIPT has come about, soon we're going to start doing it on every single patient. And many patients who (providers) would have never done an amnio(centesis) on are now going to come back with sex chromosome abnormalities flagged on their NIPT. You would have never known this because you would have never seen an abnormality on an ultrasound. And this would have never come up on their AFP (alpha-fetoprotein) testing. But now they've got some Klinefelter or whatever. […] Most of these (fetuses) do not have any visible problems on an ultrasound, so I see the baby on the ultrasound, and I go, 'she looks good to me!' And the patient goes, 'Great baby's fine! Was that test (NIPT) imaginary? Is it true? Is it not true?' […] It's interesting, we will be seeing more and more sex chromosomes problems come up […]. The big one that we always saw plenty of was Turner's because that tends to have phenotypic abnormalities on an ultrasound and it's a big cause of miscarriages too. […] But the Klinefelter and all of those, those tend to be just like really normal pregnancies, and you would have never known. But now you've got this NIPT at 12 weeks that told you this kid has potentially got a problem." (Dr. Harris Interview 2023).

Dr. Harris explains how the expected rise in SCA diagnoses during pregnancy can seem like a new 'problem' for providers and patients to navigate. With NIPT, there are more instances where a fetus' sex chromosomes can be interpreted as 'discordant' with their anatomically sexed development, disrupting the established understanding of what makes for a normal pregnancy per ultrasound. Consequently, pregnancies that doctors would have otherwise labelled 'normal,' and designated per the sex-gender binary, might now be considered 'abnormal.' However, the diagnostic implications on a to-be child's health and wellbeing remain unclear because many SCAs do not present phenotypically in-utero – as Dr. Harris notes, "this kid has *potentially* got a problem." This why SCAs remain a largely under-diagnosed medical category; aside from some severe cases, as seen with Turner's Syndrome, SCAs often do not impact a pregnancy and children can be very mildly affected during their life course, if at all (Navon 2019; "AXYS" n.d.). Indeed, a genetic counselor Rosemary, who works in a fertility clinic that provides carrier screening (which informs individuals of conditions that they are genetic carriers for), describes several of her patients having learned about their SCAs much later in life because it did not significantly impact them otherwise: **"**I've had a couple of people diagnosed with Trisomy or Monosomy X themselves […]. I had one woman who was 42 years old, never had any issues at all her whole life, needed a little extra help in elementary school with reading but that was it, and lo and behold, she had Trisomy X (Rosemary Interview 2021)." Despite their mild presentation however, genetic counselor Amy discusses how regularly identifying SCAs via NIPT complicates patientcare and reproductive decision-making:

> I've seen patients go both ways (on termination); I've seen patients terminate for sex chromosomes abnormalities. *There's a big difference between a fetus with Turner Syndrome, who has a cystic hygroma heart defect and it's likely not to survive the pregnancy versus a sort of physically typical looking ultrasound, like XYY which in general has some mild features but also most people with it are never diagnosed. And it's a more relevant question now than before, because the only people we were ever getting sex chromosomes on were people doing (diagnostic testing) procedures, and now it's a huge percent of people because NIPT is designed to pick them up.* […] So, now, we're looking at so many more fetuses' sex chromosomes abnormalities. […] Klinefelter's is a good example because structurally they're going to be normal on ultrasound, but they may have other features as they age: infertility, gynecomastia, maybe some subtle learning differences. […] I've seen everything from, 'That doesn't really sound like anything' to 'I need to terminate the pregnancy.' […] *We must be finding a lot more fetuses with intersex conditions than ever before because we're getting sex chromosome information on fetuses we historically wouldn't have. And so we're seeing that fetuses, their chromosomes and what they see on ultrasound, don't match. […] And that weirds people out a little, like, 'wait you told me from my NIPT that it was a boy and now you're telling me on ultrasound it looks like a girl?'* It's just not something that people know is a possibility. *Whereas that person, let's say with androgen insensitivity may not have actually presented with anything, and may not have been diagnosed until they were like 16, but now we're finding out when they're a fetus that they may have this condition.*" (Amy Interview 2021, *emphasis added*)

SCAs not only emerge as a more relevant diagnostic category due to NIPT's routine identification of sex chromosomes, but the diagnosis itself also implies that there is something wrong with one's pregnancy. While there are often concerns related to the future child's health needs and risks, SCAs

are also medicalized based on a limited definition of how sex and gender interrelate. As both Dr. Harris and Amy discuss, SCAs (aside from some Turner Syndrome cases) are typically so mild that they remain undiagnosed, particularly in-utero. This begs the question: do SCA diagnoses – by disrupting the conception of a necessary alignment between one's chromosomes and anatomy – entail that a pregnancy is indeed 'abnormal' or risking a to-be child's wellbeing? Even as SCAs are more frequently identified via NIPT, these diagnoses are often without a clear interpretation around whether or how severely a person might be affected; still, the mere suggestion of a diagnosis leads some parents to terminate. In other words, for many fetuses, had NIPT not flagged their sex chromosomes, there would be no evident need for a diagnosis or termination discussions – they would be considered 'normal' in their health and sex and gender presentation. For others, the issues brought on by the SCA would be addressed as they emerged, for example during puberty or fertility challenges, and the diagnosis would not necessarily shape their life from birth.

However, as Amy describes, the mere presence of a diagnosis implying an 'abnormality' puts more patients and providers in a position where they need to contend with this information. As NIPT routinely returns information about sex chromosomes, patients and providers are now asked to make reproductive and prenatal care decisions based on possible SCA diagnoses that may never materialize – and, at times ambiguity around the potential diagnosis can be stressful enough that parents elect termination. Moreover, parents can begin to question their expectations for their child's gender. As Amy discusses, patients are left confused when NIPT, having detected Y chromosomes, tells them they are having a boy while ultrasound may show female genitalia. Not only are parents unprepared for this potential finding, but they begin to question how they may relate to their child on gendered grounds; yet, without NIPT revealing a fetus' sex chromosomes, the family's life may not have been shaped as 'different' or 'abnormal' at all.

Providers, too, are not always sure of how to interpret SCA diagnoses to help patients' reproductive decision-making, which leads to further ambiguity and complications in patientcare. In several genomics conferences, genetic counselors in particular pushed back on using NIPT to routinely detect sex chromosomes. For instance, at the National Society of Genetic Counselors (NSGC) 2021 Annual Meeting, a genetic counselor presented a case underscoring how NIPT could be more harmful than useful to expecting parents with regard to SCAs. She described a pregnant mother who had pursued NIPT to learn her child's 'gender.' Much to her surprise, the screening suggested her fetus had XY chromosomes (typically male) alongside female genitalia on ultrasound. The mother declined further diagnostic testing but agreed to repeat the NIPT. A second and third NIPT predicted her pregnancy to have 47,XXY or Klinefelter Syndrome. Ultimately, she and her husband continued their pregnancy and delayed all other testing until birth. At birth, the pediatric genetics clinic confirmed their infant to have 46,X,t a translocation that the counselor clarified is "associated with a female phenotype, typical intelligence, and normal fertility." "The patient is missing her SHOX gene on her derivative X chromosome, so she may display short stature, and she does have a 25% risk of passing on the derivative X chromosome to a son, which can result in a more severe phenotype. Other than these findings, it is quite possible that she will show few to no symptoms associated with her translocation" (Genomics Conferences Ethnography 2021).

The genetic counselor went on to highlight the challenges that the parents, rather unnecessarily, had to endure given that the child would be so mildly affected. The parents "had a difficult time accepting that their daughter was going to be 'normal' with her translocation. Without NIP(T), the

patient could have lived her entire life without knowing her karyotype, but now she will have to process through this genetic difference," which has already impacted how her parents understood her gender identity as a facet of her genetics. To conclude, the counselor emphasized the need to pre-test counsel patients on the possibility of SCA findings when they elect NIPT. In another panel at the 2021 Annual Meeting for the American College of Medical Genetics and Genomics (ACMG), a genetic counselor emphasized that NIPT "should be limited to clinically severe disorders or well-defined severe phenotypes," distinguishing these from SCAs. She highlighted, "The phenotype of sex chromosome aneuploidies is variable and milder than the phenotype of the common trisomies we have been screening for" and went on to (rhetorically) ask her fellow providers, "Would there be a benefit of identifying these individuals that have a phenotype that is so mild that otherwise would do no harm?" (Genomics Conferences Ethnography 2021).

Providers, particularly genomics providers, are well-attuned to the new problem that NIPT is creating regrading SCAs and notions of gender discordance. At professional conferences, they raise important questions around which genetic differences are worth medicalizing as 'problems,' which inevitably contour how an individual experiences and develops their identity around health, normalcy, and social acceptance. The stance among many genetic counselors, those responsible for the bulk of patient genomic education, was that SCAs need not be pathologized as they tend to occlude an accurate understanding of a healthy or 'normal' pregnancy. Parents who learn about SCA diagnoses can become unduly preoccupied with their to-be child's sex chromosomes, which may be entirely clinically irrelevant in the child's life course, while relatedly worrying about their child's future gender identity as a socio-medical concern. As such, genomics providers expressed need for more intention behind prenatal diagnoses; if a genetic difference is unlikely to impact a pregnancy or a child's pressing health needs, it is perhaps not worth diagnosing at all.

However, given that NIPT exists and is widely used to screen for sex chromosomes, genetic counselors and physicians must incorporate SCAs into their prenatal care approach. Many described holding off on providing patients with an in-depth education about SCAs until and unless NIPT indicated a potential finding, at which point they could better speak to the specific genetic variation. Still, even with this tempered strategy aimed at reducing patients' anxieties, the very presence of an SCA diagnosis, no matter how mild, sets a stage where providers and patients are required to discuss sex and gender as medical concerns. It is in these moments that expectations around how an individual's sexed features ought to conform to a male/female binary, and consequently define their future gender identity as a man or woman, are sharpened. In using NIPT this way, the boundaries around what consists of a 'normal' sex and gender presentation are narrowing, with a seeming return to outdated notions that bodies determine social identities.

There are significant consequences to how NIPT is bolstering SCA diagnoses and gendered pathology, despite a lack of clinical evidence suggesting cause for medical concern. In what follows, this chapter explores three critical consequences of the ever-growing possibilities of routine genetic testing and prenatal diagnoses. Specifically, it examines the harmful social repercussions SCA diagnoses can have when they pertain to archaic conceptions of sex and gender. First, I discuss how physician providers frame SCAs as a medical concern. In an effort to uplift the attention given to SCAs, doctors and researchers pathologize a future child's gender expectations, transforming fundamentally social ideas about gendered existences into medical terms. Second, I show how the consistent push to diagnose conditions prenatally with genetic

testing leads to more extreme forms of medicalization and surveillance at the earliest timepoints in human reproduction, often with no clinical benefit. With NIPT illuminating SCAs in-utero, as opposed to when these issues *might* have presented in a person's later life, issues such as short stature and infertility increasingly medicalized and framed as prenatal problems. Lastly, I delve into how routinely diagnosing fetuses with SCAs, especially without known clinical implications, can convolute and undermine parents' reproductive decision-making as well as providers approach to equitable patientcare. A diagnosis that may never substantially impact a child's life, but that nonetheless implies abnormality just by being identified, creates undue stress and anxiety with tangible consequences for one's pregnancy. Importantly, decisions around termination, particularly as they relate to a narrowing understanding of sex and gender, hang in the balance.

Taken together, these implications illustrate how reproductive genetic technologies, when implemented uncritically, can transform socially defined phenomenon like sex and gender into medical concerns with detrimental costs for individuals and societies. The data points to a revolving door between 'objective' science and social subjectivities. As harmful essentialized views about our gendered identities are perpetuated and reinforced through technoscience, there are lasting consequences for how we collectively define 'acceptable' or desirable existences.

Pathologizing Gender Expectations in the Making of SCA Diagnoses

It's not enough that a genetic difference can be identified for a diagnosis to take hold. This difference must be considered a 'condition,' or a medical concern worthy of treatment for experts and patient communities to act upon it. Using ethnographic data from genomics conferences and interviews with providers, I show how genomics researchers and providers frame SCA-related genetic differences as deserving of attention and resources, necessitating medical interventions and participation from both the scientific community and patient families. In this process, which unfolds differently at conferences than it does within clinics, social foundations of sex and gender are transformed into biological facts. At genomics conferences, researchers uplift the need for medicalized attention toward SCAs by pathologizing non-normative sex and gender expectations; they rely on normative or cisgendered narratives of what it means to socially exist as a boy/man or girl/woman and define an individual with an SCA as potentially aberrant from these expectations. As a result, those with SCAs are seen as embodying an inappropriate gendered existence, stirring issues for themselves and their families as they move through society and life. With early interventions, however, genomics researchers emphasize that this deviation can be 'fixed' so that individuals meet normative cisgender expectations for a boy who can play rough on the playground, or a girl who doesn't appear 'too tall' among her peers. And while physician-researchers are perhaps the loudest voices in pathologizing SCAs this way, these narratives are reinforced when patients discuss SCAs as health concerns with their reproductive physicians in clinics. As interest in illuminating the SCA diagnostic category is amplified with NIPT's ubiquity, these discourses narrow social acceptability of various gendered identities and reignite stigma around sex and gender 'discordance' being an abnormal or perverse existence.

First, I rely on observations of genomics conferences to show how physician-researchers discuss SCAs among colleagues. At each conference, there were several panels dedicated to sex chromosome aneuploidies or discordant sex disorders, as researchers often termed them. Typically, physician-researchers – that is, doctors who are pioneering research around prenatal genetic conditions and possible treatments – would take turns delivering their presentations around

particular SCAs with an emphasis on the post-natal interventions they found successful. Each presentation consisted of patient case examples and took the audience (mainly other medical providers and genomics researchers) through the patient's journey from identifying an SCA in-utero via NIPT and diagnostic tests, to pursuing early interventions consisting of hormonal therapies, surgeries, and behavioral or learning supports.

A recurring pattern emerged in how physician-researchers motivated their presentations; panelists would establish the 'scientific fact' that SCAs have been largely underdiagnosed and misunderstood, but that the routine use of NIPTs shows that these conditions are far more prevalent than previously believed. A repeated statistic cited was that 1 in 200 fetuses tests positive for SCAs on NIPT results, marking quite a significant occurrence. Physician-researchers would also assert the importance of learning fetal sex as a critical moment for parent-child bonding, illustrating how social conditions around sex and gender find their way into medical landscapes. In doing so, they harnessed the growing incidence of SCAs to make the case for more routinized prenatal testing that can lead to early interventions, all of which was framed around pathologizing gender expectations for affected children and their families. Each presenter sought to establish their approach to SCAs as the 'best' practice (influencing best practice guidelines is a common discussion at such medical conferences), while also garnering more attention and funding for their line of research on SCA diagnoses and treatments. In this process, 'discordance' between one's sex chromosomes and anatomical sexed features, with assumed consequences for one's lived gender identity, was pathologized into a condition warranting medical treatment to align these children with normative binary sex and cisgender expectations. The examples below illustrate how prominent physician-researchers bolstered their perspectives around routinely using NIPT to test for SCAs and following through with early interventions.

At the 2021 ACMG panel titled "Sex Chromosome Trisomies – To Screen or Not to Screen?" (wherein all the physician-researcher presenters concluded 'to screen'), the first physician-researcher presenter aimed to convince audiences of the need for more routine testing of SCAs. She began by describing how SCAs have been long misdiagnosed, noting that affected individuals have "mild or absent phenotypical characteristics" and "non-specific symptoms." She discussed a wide range of intellectual, behavioral, and physical symptoms associated with SCAs, including language-based learning difficulties (e.g., dyslexia, dysgraphia), executive dysfunction, delayed complex motor skills and developmental dyspraxia, tall stature, anxiety and ADHD, and hypotonia (decreased muscle tone). Ultimately, however, she noted that these challenges are typically resolved with time and extant supports (e.g., behavioral and hormonal therapies). The physician-researcher concluded "the kids go onto lead pretty normal lives." (Genomics Conferences Ethnography 2021).

What she subsequently framed as less than normal, however, were the sex and gender presentations that these children may experience. She relied on examples of infant patients who presented with Klinefelter Syndrome (47,XXY). Displaying images of these boys' "decreased phallic size," the researcher went onto describe how critical treatments would need to focus on "helping maleness to develop" or support "male typical behavior" in an effort to "help masculinization." The presentation invoked concepts of normative sex and gender as the treatment objective, as these boys were pathologized for sex chromosomes and genitalia that, presumably, did not correspond with how boys are *supposed* to look, behave, or socialize. The researcher concluded by discussing

how prenatal diagnosis of SCAs would enable much-needed preparation for a range of possible issues; parents and providers could gather information, find support groups, connect with specialists (e.g., geneticists, counselors, endocrinologists), and intervene in challenges as early as possible to support "mother-infant bonding" around raising a child of a particular gender. "If things are done *correctly*," she stated, "these boys can go into kindergarten with no additional support needs." Explicitly, she establishes that there is a 'correct' way to raise individuals with Klinefelter Syndrome so that they fit a normative gender binary. (Genomics Conferences Ethnography 2021, *emphasis added*).

This researcher was certainly not the only one to pathologize gender expectations for those with Klinefelter Syndrome. In another similar panel at the 2021 Philadelphia Prenatal Conference, which doctors described as a major reproductive medicine gathering with "big name" presenters, a geneticist discussed how boys with Klinefelter "can be at high risk for jail or breaking the law […]." She portrayed these boys as possibly more aggressive stating, "there can be school problems or legal concerns," unearthing debunked notions of a 'criminal chromosome' and fueling the scientific imaginary around genetic essentialism.[42] On the other end of the spectrum, she described that some boys with Klinefelter can be "shy, sensitive, and withdrawn […], making them susceptible to bullying." For girls with Klinefelter, the presenter focused on problems with their fertility and tallness. Where being shy and sensitive was an unacceptable gendered behavior or personality for a boy, for girls, being 'too tall' or infertile was inappropriately unfeminine. Rather than expand our notions around gender to diminish the harms of social rejection and bullying, the physician-researcher positioned shyness and tallness as issues necessitating medical treatment that would align these children with normative gendered expectations. As such, sex and gender go from being social concepts to becoming medical facts with diagnostic implications. (Genomics Conferences Ethnography 2021).

The remainder of the talk focused on how "early treatment" with hormone replacements and "booster shots" "is needed to mitigate the effects of the additional X" for boys with Klinefelter – to cure them of their femininity. The physician-researcher discussed at length how the "development of maleness" can be achieved with hormones, androgens, and various behavioral and physical therapies. She paid significant attention to muscle development and penis size for these boys, asserting that physical features would be critical for them to appear more like their 'normal' cisgendered male peers. To emphasize the importance of early diagnosis and treatment, the presenter described that "a five-year-old boy" would be considered "wimpy or baby-ish" if their community did not know about their Klinefelter condition; but with a prenatal diagnosis, "it changes how their parents treat them" for an "optimal outcome." She frames her proposed interventions as "giving parents hope" and helping them "avoid a shock" when it comes to their expectations for raising either a boy or a girl. "If parents are guided and supported, *these children can be indiscernible in the population and go on to live normal lives* […]. (They can) lead successful and independent lives with very little mental health and learning supports." Not only are gendered expectations pathologized to garner significance for SCA diagnoses and treatments, but these agendas are centered on how affected individuals may be socially perceived on gendered

[42] Similar to Jacob's Syndrome (47,XYY), Klinefelter Syndrome (47,XXY) has also been inaccurately linked to aggressive behaviors, violent tendencies, and criminality (Stochholm et al. 2012). In both cases, essentializing such behaviors and outcomes to one's genetic composition or differences is not only scientifically unsound, but overly deterministic and ignorant of the important effects that social structures have on one's life.

grounds (as opposed to clinically meaningful interventions for their health and wellbeing). The boundaries around 'normal' or socially acceptable sex and gender identities are increasingly narrowed in the process. (Genomics Conferences Ethnography 2021, *emphasis added*).

This focus on parental and societal expectations of a child with an SCA seemed to take precedence over the child's realized medical needs. Despite many children with SCAs not actually requiring medicalized interventions for health issues, physician-researchers almost exploited parents' anxieties around their child's gender and genetic difference in order to motivate the *need* for treatments. For example, another physician-researcher at the 2021 ACMG meeting shaped their talk entirely around parenting expectations for raising a child who did not confirm to a cis sex-gender alignment. Notably, the presentation did not draw on accounts from parents describing such anxieties, but rather assumed these to be concerns based on a medicalized view of sex chromosomes and tropes of masculinity and femininity. Again, the researcher used the case of Klinefelter, stating that this diagnosis is becoming one of the most common prenatal conditions with 1 in 600 births being affected. He described boys with Klinefelter as having "reserved social skills" and "quiet, docile personalities," which he characterized as inappropriate masculine development. The early interventions proposed revolved around a boy's "mini puberty," an "important stage in life of all boys" at merely 3-6 months of age. Describing gender as a part of an infant boy's brain (despite well-established critiques of such neuroscientific approaches to gender, see Jordan-Young and Rumiati 2021), this researcher emphasized the need to study and address SCAs using early hormonal therapies that would target 'normative' brain development around gender (especially regarding communication and language) to ensure boys undergo their mini puberty and resemble hetero-typical masculinity. (Genomics Conferences Ethnography 2021)

The presenter provided specific patient examples to illustrate the supposed urgency around early interventions that would ensure an infant boy's mini puberty. He described boys with Klinefelter as "falling further behind in terms of strength, ability, and different types of sports," highlighting how expectations for masculinity are often predicated on physicality and sports, where an inability (or disinterest) to perform this way is considered a gendered aberration. In particular, the researcher focused on one child who had inguinal (undescended) testes, showing graphic images of this child's body to portray their sex as a problem that would perhaps shock and disgust audiences. After showing the images and remarking on the child's deviance from binary sex and cisgender expectations, the physician-researcher stated, almost as a given, "it would have been much better to have known this in utero than have to deal with the *embarrassment of the gender*." He depicts sex-gender 'discordance' as a humiliation to parents who become unsure of how to understand their child's gender per the differences in their sex chromosomes and physical features. The researcher then asserts that sex classification at birth is an important milestone: "plans need to be in place at birth to assign the *proper* sex." He suggests that there is an objectively correct sex that individuals need to be assigned, per a sex-gender binary, in order to transition a child's identity from being discordant to concordant with societal expectations of their masculinity or femininity. He situates this approach to sex assignment as a precondition for one's cisgender identity and life course, repeatedly stating that "sex assigned at birth informs rearing" and that SCAs "generate confusion about sex and rearing." Throughout, the target remains parental relationships and societal reactions to a child's gender identity, rather than a medical concern posing harms to the child's health and wellbeing. 'Abnormality' thus becomes less about the child's medical needs

than about social prejudices around their gender, illustrating how scientific classifications are subjectively constructed. (Genomics Conferences Ethnography 2021, *emphasis added*)

Before ending the talk, the physician-researcher took a few questions, addressing smaller yet critical points. For example, while acknowledging the debates around when to offer fertility preservation to children with Klinefelter, he emphasized that fertility interventions are an important aspect of these individuals' "normalcy." Not only was fertility seen as essential to one's gender identity, but the researcher also highlighted parents' desire for grandchildren as a key motivation for early fertility treatments, illustrating how reproductive capabilities become central to medicalizing male- and femaleness. Throughout this presentation, there was a consistent focus on making boys into men, girls into women, and ensuring masculinity and femininity could be biologically, rather than socially, defined with prenatal testing and interventions. (Genomics Conferences Ethnography 2021).

While many talks focused on the appropriate presentation of maleness, using Klinefelter Syndrome as an example, a few delved into notions of acceptable femininity. Here, physician-researchers typically drew on examples of young girls with Turner Syndrome (Monosomy X). For example, at the 2021 Philadelphia Prenatal Conference, a geneticist and then-president of the ACMG delivered a presentation calling for increased use of NIPT to identify a fetus' sex chromosomes and enable "correct" sex assignment at birth followed by early interventions for those with SCAs. This presentation focused on an example of two sisters, one with Turner Syndrome and one with typical XX chromosomes. He continually pointed the audience's to the affected sister's shorter height and build as a less-than-feminine stature. Various images of the sisters were displayed side-by-side, with black boxes obscuring their eyes, so that their identities could be somehow concealed while the audience observed the gendered anomalies of one sister compared to the other. The geneticist heralded this case as a success – his growth hormones treatment plan enabled the sister with Turner Syndrome to "reach the first percentile" for height. He contrasted this with another case, where a couple declined further testing following an NIPT suggesting SCAs; they saw their twins appearing healthy and 'normal' on ultrasound, and thus did not feel the need to medicalize their pregnancy. Even though the parents were delighted at the birth of a twin boy and girl, this geneticist considered this case a failure because the family declined further prenatal testing and diagnosis of an SCA. At times, it seemed concerns around children having SCAs and 'gender discordance' were more foregrounded in physician-researchers' minds than they were for parents. (Genomics Conferences Ethnography 2021)

Concluding his talk, the geneticist reiterated "it's important to document fetal sex. *Failure could have grave consequences for the newborn or young adult*." However, the direness of these consequences for patient families who forwent or resisted SCA diagnosis weren't readily apparent through his presentation – indeed, the parents of the twin boy and girl did not seem perturbed by their children's outcome. Nevertheless, the researcher emphasized his perspective on routinizing NIPT: "[NIPT] is a huge opportunity to identify sex discordance prenatally […]. By doing so we start that early testing once the child is born and begin any treatments. […]. We need to identify the biologic sex of a baby, so we can eliminate anxiety that comes with birth. *Parents always want to know 'what is my child? male or female?' We know that has important psychological effects for parents. Early testing can get them prepared accordingly for how they will treat their child.* How the child will actually be treated is another discussion, but identifying biologic sex before birth is

really important." Not only is gender pathologized based on tropes of what it means to exist in society as a boy/man or girl/woman, but parental anxieties (and social perceptions) are prioritized as a justification for medical interventions. While notions around appropriate or acceptable sexed and gendered existences emerge as powerful currents undergirding the fabric of our intimate and social relationships (West and Zimmerman 1987), pathologizing these identities in the making of SCA diagnoses restricts our understanding of meaningful and diverse lives to that which is scientifically imposed and biologically determined. (Genomics Conferences Ethnography 2021).

To a non-medical observer, particularly a sociologist, it was hard to miss that these prenatal genomics panels were fundamentally about sex and gender assumptions, situating gender as a necessarily embodied identity predicated on a binary understanding of sex. None of the physician-researcher panelists considered themselves to be commenting on gender; almost all began with a disclaimer stating their talk was *not* about gender, that they understood gender to be a social construct. To them, the discussion around testing and treating SCAs was objectively about 'discordant sex' as a medical condition, with dissonance around how gender was being pathologized in the process. Seemingly, the physician-researchers did not realize how their justifications for amplifying attention toward SCAs were built on a narrow understanding of binary sex and cisgender expectations. When one geneticist at the 2021 ACMG meeting was asked "Is some part of testosterone for strength treatment for boys to strengthen male gender identity?" they responded: "The boy has a deficiency of testosterone which affects female-based disorders and his health and well-being. *If (the patients) want to make this a part of their gender, that's their decision*." The geneticist described that testosterone has impacts on bone health, physical fitness, lupus, muscle tone and strength, academics, and athleticism, in an effort to contextualize this SCA as a health (rather than gendered) issue. Still, she ended her comment with an undeniable return to pathologizing gender to uplift the need for medicalized SCA treatments: "*If you're a boy and you can't play on the playground because you're not strong enough, it's devastating*." Although she was pathologizing particular gendered presentations, to her, this was largely obscured in the face of assumed scientific facts and biological determinations. Ultimately, the 'problem' for boys with lower testosterone stemmed from a sense of shame on the playground among their peers, who they presumably would not fit in with due to their lacking masculinity. As social expectations cascade into medical frontiers, a lack of alignment between one's sex chromosomes, genitalia and gonads, and gender identity is increasingly considered an 'abnormal' and unwanted existence. (Genomics Conferences Ethnography 2021, *emphasis added*).

Next, using interviews with practicing physicians, I delve into how this gendered pathology unfolds within clinics during physician-patient consultations. In contrast to physician-researchers at genomics conferences, doctors in clinics did not emphasize the need for NIPT to routinely detect sex chromosomes nor the urgency around medicalized SCA treatments. In clinics, the SCA-related discussions between doctors and prenatal patients appeared more centered on the pragmatic medical management needs of each pregnancy. However, in this process, as they increasingly employed tools like NIPT and ultrasound to classify fetus' per a sex binary, these providers (perhaps inadvertently) positioned associations between sex and gender as a necessary condition for normalcy. Accordingly, discourses pathologizing gendered continued within the clinic among patients and reproductive physicians, further reinforcing the notion that biological sexed features determine gender, and that any aberrations would lead to unfulfilling lives.

The reproductive physicians interviewed, many of whom were maternal fetal medicine specialists with expertise in genomics, described that sex chromosome variations typically did not concern them. They saw NIPT's value being its ability to non-invasively screen for more severe conditions like Down, Edward, and Patau Syndromes, which have consequences for pregnancy management and immediate health interventions for a child (if the fetus makes it to term, which is often not the case with Edward and Patau Syndromes). As Dr. Li stated, "(SCAs) are not really a medical concern. […]. I would say in general, genetic (NIPT) screening is for picking up Down Syndrome risk because that is the most common genetic abnormality (Dr. Li Interview 2021)." SCAs, on the other hand, were described as milder and far less impactful for an individual's health. Unless an ultrasound pointed to structural issues within the fetus, perhaps associated with an SCA, doctors in clinic were not fixated on using prenatal genetic testing to learn a fetus' sex chromosomes because this information would not change a patient's prenatal clinical care and any phenotypical concerns would be mild and addressable as needed during one's later life course. Dr. Janowski, a maternal fetal medicine expert with over 14 years of experience working at a large community hospital, explains her perspective on SCAs and fetal sex chromosomes:

> *No (SCAs) are not medically concerning. Do you need to know the sex chromosomes of your baby before it's born in order for it to have a healthy life? No.* Sex chromosomes, generally, are not determined until after the baby is born, or never. I don't know what my sex chromosomes are. I'm assuming it's XX because I made another baby, but I don't really know, I didn't have my carrier type checked. […] *You don't need that information to parent your child. Then if something interesting or weird happens to your kid, you'll get some testing if you think that would be helpful at that point.* (Dr. Janowski Interview 2021, *emphasis added*)

For doctors managing patients' pregnancy care as their main responsibility, information around sex chromosomes does not usually affect prenatal clinical decisions. Further, as Dr. Janowski discusses, knowing an individual's sex chromosomes is neither standard practice for health management nor medically meaningful for patients' parenting. As such, doctors in clinic did not find much value in having sex chromosome information prenatally via NIPT, unless ultrasound indicators suggested other possible complications. These doctors also more conscientiously appreciated the complexity and ambiguity within SCA diagnoses, in that it was not always clear whether or how a child may be affected, which in turn complicated patients' reproductive decision-making. There was a greater understanding among physicians in clinics that not all patients wanted to medicalize sex chromosome variations, and indeed most need not be medicalized for children to be born healthy and parents to find joy in their new families. Dr. Scott, who has been in the field for over 45 years, reflects on an instance where her team seemed to be more concerned about an SCA diagnosis than the pregnant patient herself, regretting how it impacted the patient:

> I had one patient […], her second baby was possible Turner's Mosaic by (NIPT) screening test. She didn't even get a diagnostic test. But when she was showed the ultrasound or at visits, all (the providers) wanted to talk about was the Turner's and she knew her baby was normal. It turned out she was right. And she totally got alienated because of bias on our part. […] I mean we're learning that female babies with Turner Syndrome, they can have children. They can do IVF and use those eggs. And some aren't disabled intellectually. So

> we're also learning that our gloom and doom, because the disabled aren't well researched, sometimes is overreaction. (Dr. Scott Interview 2021)

Dr. Scott recognized a moment where an SCA did not need medicalization. She went on to discuss the dearth of research around disabled communities, and how this impacts the way providers handle genetic differences. While this particular patient did not ultimately have a child with a concerning presentation of Turner Syndrome, there are instances where doctors do need to counsel patients whose fetus' show more challenging SCA presentations. In rare cases, SCAs can be quite severe and require significant medical attention. At the 2021 NSGC meeting, a genetic counselor presented a case of a 17-month-old infant with female genitalia who had a rare deletion of their DMRT1 gene, leading to a 46,XY sex chromosome variation.[43] This infant needed prompt medical care, as there were concerns around 'complete gonadal dysgenesis' (absence of ovarian tissue) associated with a high risk of developing gonadoblastoma, which is a tumor in the gonads that can turn cancerous if not treated. Other presenters also discussed more severe cases of Turner Syndrome, which can have implications for fetal mortality and may require coordinated care among maternal fetal medicine, cardiology, endocrinology, genetics, and psychiatry specialists (Genomics Conferences Ethnography 2021). In these cases, doctors emphasized discussing the clinical aspects of an SCA with patients (i.e., implications for fetal organ development and function, typically evident via ultrasound) to keep the conversation as 'objective' or focused on medical management for health needs as possible. Some doctors, like Dr. Khoury, very intentionally steered these discussions away from gender:

> For things like Turner Syndrome, XO, (patients) questions are like, 'what does that mean? What is an XO?' I think it's really easy when you describe it to stay scientific and it's not about gender, it's just sex chromosomes and we keep it really just about sex chromosomes and the implications to the heart and the implications to the height and the phenotype. (Dr. Khoury Interview 2021)

Most physicians described their clinical approach similarly – sticking to the phenotypical presentations that could be objectively linked to the SCA. Realizing that questions around sex and gender could also be important to patients, these doctors would rely on a gamut of resources, including genetic counselors, specialists, and patient literature to direct families toward further support. As such, reproductive physicians in clinics tended to concentrate more on managing the structural issues a fetus may present, as these had bearings for more involved prenatal clinical care, while leaving more in-depth counseling around sex and gender to other providers. Dr. Leighton, who has been involved in prenatal care and research for almost 20 years, describes her reliance on genetic counselors and other specialists:

> For the sex chromosomes, if (patients') NIPT comes back worrisome for that, *(patients) usually go straight to genetic counselors*. […] Definitely the hardest decision (with regard

[43] Swyer Syndrome (46,XY) is characterized by undeveloped gonads (i.e., testicles or ovaries). Individuals with Swyer Syndrome usually appear phenotypically female, with functioning female genitalia and reproductive structures. However, because their ovaries have not developed, they do not naturally produce sex-related hormones or experience puberty and may face other challenges such as gonadoblastoma (tumor in the gonads). The severity of this condition is variable, and for some may not be detected until they face issues with puberty. Typically, treatment consists of hormone replacement therapy. ("Swyer" 2019, "Swyer" 2022)

> to termination) is when the brain is abnormal but subtly so, meaning (the brain is) missing a structure, (but it's) survivable, (has a) spectrum of outcomes, (and) normal testing. That's a hard situation. Some (patients) terminate, some don't. *The way we help folks through that is we have them meet with pediatric neurology. We have them meet with a variety of people to help them through that decision.* (Dr. Leighton Interview, *emphasis added*).

Similarly, Dr. Liang, who has practiced in a private obstetrics and gynecology setting for 32 years, describes the robust resources he has turned patients with prenatal SCA findings to over the years:

> Most likely it would be either the Monosomy X or a XXY or XYY that would turn up, statistically. In general, we have a brochure to hand the patients that have something like Monosomy X. We look up either online literature or support groups for parents, or even for patients with that condition. Depending on the situation, I could make a referral […] for the genetics counselor or for the medical geneticist. *But (SCA) doesn't really concern me. As long as the pregnancy got as far as the second trimester, Monosomy X is likely to survive and do fine. I'd mainly want to make sure there's no congenital heart disease involved.* And so we've got a fetal echocardiogram for that. *Everything else is generally fine, it doesn't impair perinatal outcome or effect time or place of delivery or route of delivery.* (Dr. Liang Interview 2021, *emphasis added*).

Dr. Liang highlights, once again, how SCAs are typically not a medical concern for him as a prenatal practitioner because they typically do not impact fetus' health in-utero nor the plans for delivery and clinical care a fetus would need at birth. While he is attuned to particular structural differences, such as within the fetus' heart, he generally discusses SCAs as uncomplicated pregnancies that benefit from a range of social supports.

However, even as doctors in clinic tend to focus on pragmatic clinical management and fetuses' structural issues, they also pay significant attention to genitalia on ultrasound. Here, 'objective' clinical management often resembles gendered pathology, as there is amplified medicalized attention toward classifying a fetus per binary sex in order to establish its normalcy. Although genitalia are an important part of one's anatomical development, research has long shown that genitalia do not conform to a male/female binary, and that there are at least five sexes that individuals' genitalia may represent (Fausto-Sterling 1993). However, this broader understanding of sex outside a binary classification did not translate to doctors' clinical practice. Visualizing genitalia on ultrasound, to make a call around whether the fetus presented as typically male or female, was considered a medical standard and non-negotiable among doctors in clinic:

> There are people who don't want to know the sex which is fine. They still get the NIPT they just don't look at it. […] On the anatomy scan I'm like, 'I'm just going to look here, you can pull your eyes away, *but we still have to look to make sure that it looks normal, of one sex, and that it's not ambiguous*,' and then we move on (Dr. Li Interview 2021, *emphasis added*).

While doctors in clinic did not find SCAs to be medically concerning, the idea of a pregnancy being 'normal' was predicated on a fetus developing genitalia that could readily be classified male or female (in contrast to 'abnormal' intersex genitalia). The focus on fetal genitalia broaches

patient-provider conversations around what the to-be child's eventual sex and gender identity may be, with sex and gender being situated in a narrow and unrepresentative cis-conforming binary. Further, although many doctors acknowledged that findings of ambiguous genitalia on ultrasound were distinct from one's gender identity, they did recognize how the relationship between sexed features (i.e., genitalia) may prompt parents to consider gender expectations for their to-be child. Dr. Leighton and Dr. Li, who is in her fifth year of practice, discuss these critical moments wherein questions around sex and gender become a part of complex reproductive decision-making:

> The other tricky thing is when there's ambiguous genitalia. And that is that is tricky because we're not great at making that call on ultrasound, particularly early on. We get better once folks are probably at 20-22 weeks, but it's a hard call to make on ultrasound. […] It's easier now to deal with than it used to be 10 or 15 years ago, because now folks have a little better sense of gender identity issues. […] So now the level of cultural understanding is better than it was just 15 years ago. But most folks don't really know much about ambiguous genitalia at all. That is definitely a situation where I'm usually saying, 'Hey folks, this catches my eye on ultrasound. But ultrasound is not great about picking this up. So I really want you to get a second opinion.' Yeah, and usually that second opinion will be through the fetal treatment center. And what I tell them is, 'For these really ambiguous things that can have a big impact it's important to get lots of opinions and specialty opinions.' (Dr. Leighton Interview 2021).

> There was a recent patient who we saw who did IVF (in vitro fertilization), but then didn't do PGT (preimplantation genetic testing on embryos), so they didn't know the sex. And then had a normal NIPT with XY. But then along the pregnancy, at the anatomy scan, (the fetus) had ambiguous genitalia. It was not a male fetus, it had clitoromegaly (enlarged clitoris). (The patient) was counseled that most likely there was some androgen insensitivity syndrome, and she was offered genetic counseling. The MFM did the amnio(centesis) and it did come back XY. But she actually decided to terminate the pregnancy, even before seeing genetic counseling. […]. (The patients) were very concerned; it was IVF with an egg donor, so like they were very concerned about all the things with androgen insensitivity syndrome especially the infertility and some intellectual delay. […] There was another patient with a similar situation except it was a reverse. NIPT showed XX but then on ultrasound it looked like male genitalia. But I don't know, it wasn't the most obvious, it was just ambiguous. And then she had a normal amnio. She continued the pregnancy, and it ended up being just a baby girl, female sex in the end. (Dr. Li Interview 2021)

These accounts show how the clinical management of fetal genitalia, as it relates to SCAs, can be incredibly complicated for patients and providers to navigate. It's not always clear how a to-be child's health and wellbeing may be impacted, and based on parents' individual concerns, genitalia and sex chromosomes can be more medicalized in some cases than others. No matter how 'objectively' a doctor in clinic attempts to center discussions around sex chromosomes and genitalia, prenatal technologies like NIPT and ultrasound render sex and gender more visible and socio-medically relevant during pregnancy. And while doctors in clinic may not partake in gender pathologizing as explicitly as their physician-researcher counterparts in conference settings, sex and gender nevertheless emerge as consequential discussions with patients. In these moments,

knowing sex chromosomes prenatally, regardless of any realized health implications for a to-be child, engenders patient-provider conversations that fuel ideas around binary sex and biologically determined gender, and in some cases leads to termination decisions. NIPT's routine use not only keeps discourses medicalizing binary sex alive, defining 'normalcy' accordingly, but also sharpens boundaries around socially acceptable gendered existences as necessarily embodied. Along these lines, Dr. Das illustrates an encounter with patients who learned their fetus had an SCA via NIPT:

> (The pregnant patient) thought about termination. *She thought about what other people were going to perceive the kid like, and I think that was the big thing - the social aspect of how the newborn was going to be perceived as* […] I think it was XYY. It was either XYY or XXY (referring to Jacobs and Klinefelter Syndromes, respectively); one of them is where there have been more studies on patients who have a lower intelligence level and are predisposed to lower socioeconomic statuses. Studies originally thought that they were a little bit more hostile and more aggressive. But then it was later thought that most of their actions were attributed more to just being lower socio economic and less from aggressiveness. *So, I talked to her a little more about that, and that most of the times there isn't a phenotype that is displayed that would make (the child) look any different. Ultimately it was a talk that centered around the social aspect of (SCAs), about what other people are going to think.* They got more of the genetics aspect and the behavioral stuff from the genetic counselor. (With me), it was more like, 'we feel really stressed, *we have a couple of normal babies at home. And how is this one going to fit into the family, and how about socially, how are we going to cope with having other people know about it?*' (Dr. Das Interview 2021, *emphasis added*).

When presented with SCAs, doctors report that some parents worry about how their child will fit into broader society. If they cannot be classified as a boy/man or girl/woman based on their biological features, what are they? And will the child's supposed gender anomaly be a source of shame among friends and family? Providers like Dr. Das described that being able to conceptualize their child's gender identity as 'normal' – that is, reflective of their sex chromosomes and genitalia – was important for some parents to situate their expectations around raising a particular individual. Even as he reassured the patients that their child would not present with an 'abnormal' or different phenotype compared to individuals with XX or XY sex chromosomes, the very identification of a sex chromosome variance unraveled a discussion pathologizing sex and gender. As routine NIPT makes such patient-provider conversations about SCAs more commonplace in prenatal care, there is a consistent harkening back to gender expectations being biologically determined; even as doctors may push back on gendered pathology, these ideas continue to circulate among patients, providers, and medicalized spaces.

In Dr. Das' example, we also see how discredited studies (e.g., 'criminal chromosomes') continually resurface as SCAs become more visible in prenatal care. Other doctors described similar instances, where debunked science influenced how expecting parents thought of their potential child with an SCA: "The XYY (Jacobs Syndrome) is a problem in terms of counseling for the family. Before they would be like 'oh, that means my kids going to go to jail and have all those issues.' And we're like, 'no, not necessarily. Your child may be totally normal, and you'll never know. Probably someone walking in here has XYY, and you'll never know (Dr. Khoury Interview 2021)." Despite scientific research establishing that certain SCAs have been historically

misunderstood, there is a power behind pathology as medical diagnostic categories become institutionalized as social facts.

While doctors in clinic and physician-researchers at genomics conferences play different roles in medicalizing SCAs, they each participate in pathologizing sex and gender expectations in the making of this diagnostic category. Physician-researchers at conferences present SCAs in far more dire medicalized terms compared to doctors in clinic. As NIPT more routinely reveals sex chromosomes during pregnancy, physician-researchers have a heightened interest in employing this technology toward igniting greater significance (and, importantly, funding) for their research agendas focused on prenatally diagnosing and treating infants with SCAs. To garner attention and resources for their work, these physician-researchers intentionally frame SCAs as a problem of gendered expectations, instilling a sense of fear or shame in parents whose children may have SCAs. Moreover, their narratives justifying the need for medicalizing infants with SCAs are predicated on the notion that discordancy between sexed features, chromosomes, and gender will result the child being a social deviant because of their inappropriate presentation of masculinity or femininity. In this process, physician-researchers transform sex and gender from fundamentally social ideas into biologically determined and medically relevant 'facts.'

On the other hand, in clinics, doctors tend to focus more on patients' prenatal clinical needs, and often work toward comforting patients who are faced with SCA diagnoses, insisting that sex chromosome variations will not negatively impact their children's health and wellbeing. Many of these doctors take on the work of clarifying how sex chromosomes do not implicate gender identities and dispel myths around 'criminal chromosome' assumptions. Still, their practices around using technologies like ultrasound and NIPT to classify fetus' normality per their genitalia and sex chromosomes creates the opportunity for patient-provider discussions that center on how sex-gender 'misalignment' can be a social and medical concern. That these conversations take place more frequently in medical spaces, given how NIPT is making SCAs increasingly visible, reinforces regressive ideas of sex as a binary and gender as rooted in the body.

Gender pathologizing processes among clinic doctors and physician-researchers co-exist and reinforce one another. Together, they lend scientific authority to inaccurate and harmful conceptions of sex as an immutable binary and gender as necessarily conforming to this embodiment. By framing subjective gender expectations as objective medical concerns – thus defining 'normalcy' as heterotypical presentations of masculinity and femininity – these co-productive narratives among physician-researchers, clinic physicians, and patient communities perpetuate a narrow understanding of the gender identities individuals can and *should* represent.

There are also other significant and tangible consequences to augmenting the SCA diagnostic category via gendered pathology. As experts routinely use NIPT to detect sex chromosomes, the attention toward SCAs as a medically meaningful diagnoses becomes more widespread. Consequently, more biomedical research funds are poured into medicalizing SCAs as worthy of prenatal detection and early interventions, bolstering the idea that sex-gender discordancy and non-normative gendered existences are socio-medical problems or abnormalities worth eliminating. As early as 2013 (just two years after NIPT was released as a consumer technology), Genomic Health, a company that develops and commercializes reproductive genetic tests, received US$100 million from investors to work on prenatal testing for Turner Syndrome. In 2015, Natera, a genomics

company that develops NIPT technologies to test for chromosomal conditions including SCAs, raised US$200 million from investors. These trends in biomedical investments carry onto the global scale, as numerous reports predict that the global market around testing and treating SCAs will be worth over US$10 billion by 2032 ("Genomics" 2022, "Non Invasive" 2019, "Non Invasive" 2022).

The substantial resources poured into genomic technologies that medicalize SCAs indicate a growing trend toward considering these conditions pathological. They demonstrate how the narratives around aberrant sex and gender presentations take hold as 'objective' medical problems worthy of immense attention and funding. Transforming reductive social conceptions around sex and gender into medical diagnoses has lasting consequences for how individuals view themselves, accept and respect one another, and define which existences are considered meaningful. By using medical technologies and pathology to restrict the types of sexed and gendered lives considered appropriate or acceptable, there is a perpetual shrinking and sharpening of the boundaries around 'normalcy' with harmful costs for the many who exist outside these confines.

Medicalizing More, Medicalizing Earlier

As SCAs are more frequently detected and diagnosed with NIPT, doctors and patients also participate in more interactions that medicalize biophysical traits which otherwise would have remained un-pathologized and medically irrelevant at the prenatal stage (or, perhaps for the entirety of one's life course). Providers highlighted that patients often seem concerned about their future child's potential infertility and height as characteristics associated with certain SCAs. Infertility is linked to several SCAs but, today, is treatable with well-established interventions. Ordinarily, had a person not been aware of their sex chromosomes (as was the case before NIPT's routine use), they may only have learned about fertility challenges later in life, at which point they could seek relevant interventions or alternative approaches to family-building. However, with NIPT revealing SCAs and possible infertility in-utero, this issue becomes medicalized much earlier in an individual's life course – at their fetal stage, rather than perhaps decades later.

Likewise, height (or stature) is another biophysical trait that becomes medicalized much earlier in an individual's life course, as 'unusual' shortness or tallness can be associated with various SCAs. Concerns around height may never have been medicalized at all without the revelation of an SCA, given that it is unclear to what extent stature differences impact one's health or wellbeing (Coste et al. 2012, Horrom 2022). Where infertility may eventually have medical implications, height is often discussed more as a social desirability concern. Harnessing interviews with genetic counselors and physicians, where providers illustrate their discussions with patients, I show how these characteristics are medicalized as challenges to the gendered expectations parents have of their children. By positioning SCAs as a diagnosis category of concern, if only because a genetic difference has been identified, NIPT use leads to more instances where individuals can be considered 'abnormal' with increasing constraints around what life experiences seem meaningful.

In this process, the medical gaze is evermore expanded, with fetuses increasingly surveilled and pathologized based on supposedly medical markers. This heightened medical gaze has notable consequences, as considerations around biophysical traits – regardless of whether they impact a fetus' immediate health or pose substantial life challenges – do factor into parents' decisions around terminating or continuing pregnancies with SCAs. As Amy, a genetic counselor, sees it:

"Some people are kind of like, "all right, so she may be taller and might have some slight trouble in school, but otherwise she has normal fertility" and other people are really weirded out by it. […] We just try to give a balanced view […] and then patients have to decide if that's something they can live with or not (Amy Interview 2021)." Increased medicalization not only impact reproductive decisions, but providers are also keenly aware of their role in influencing these choices; as Amy notes, parents need to make these decisions based on their own priorities and values. And, even if a pregnancy is continued, the to-be child may now be considered 'abnormal,' having to process this sociomedical identity through their life course, where the SCA diagnosis may have otherwise had very little impact on their lives had it not been illuminated prenatally.

My critiques around traits like height or infertility being medicalized prenatally are not situated in a social, ethical, or political position around abortion. The issue here is not about whether one should be able to terminate a pregnancy for these reasons, or any for that matter. Pregnant people need wide and safe access to abortions, regardless of their reasoning. Rather, the concerns presented related to how uncritical increased use of genetic technologies to diagnose conditions, accompanied by practices which pathologize the identified differences, establishes diverse biophysical traits and resulting existences as less desirable than others. This research shows that our societal values around reproduction and worthwhile existences are being influenced and constrained by technologies that create new diagnostic and patient groups based on often mild differences, increasingly sharpening the boundaries between normal and pathological. In the example below, Dr. Banerjee describes her experiences with expecting parents who have learned about their fetus' SCA via NIPT and consider termination:

> Turner Syndrome, for example, there's such a spectrum of how it can present and so for some patients *if there's not a cardiac abnormality and then we're really saying, 'hey this is probably going to be like a stature issue and infertility*.' But there's such a spectrum. Different people have different ways of thinking about that. I think it's a difficult thing; *as a parent, you basically are making a choice for your future children, way later in their life that they're going to have infertility issues.* Some people feel like that's something that's very easy to overcome and then for other people, they really think about it like 'I'm potentially agreeing to a life that is hard and what if that's not something that my child would want to go through?' *Some people will be like, 'well height is a really big deal for boys, and I just don't want to like make my child's life harder.'* […] For me, the (termination) decision point is whether something is like lifelong disability, chronic medical illness. That's not a life I want for my child. But these other things (SCAs) are things that are treatable and easy. […]. (Dr. Banerjee Interview 2021, *emphasis added*)

While Dr. Banerjee clarifies her personal position on SCAs, noting she would not terminate for reasons of height or infertility, she explains how parents think about such concerns as both medical and social challenges for their child to-be. This sentiment was echoed in physician-researchers conference presentations, where several noted that parents' concerns with their future child's infertility stemmed from a desire to eventually have grandchildren; reproductive capacities are a feature of normative gendered expectations. There is a recurring question around what type of life one chooses for their child (and, by extension, themselves), and whether 'abnormal' height or infertility are enough to compromise this existence. The future individual's fertility, in particular, becomes a question much earlier because parents can access this information prenatally and are

able to make reproductive decisions based on it. Importantly, making infertility medically relevant at the prenatal stage is unique to genetic technologies like NIPT that reveal sex chromosomes and illuminate SCAs, despite the absence of other medical markers. Even if parents might have otherwise not been concerned with their future child's infertility, knowing that this challenge may exist – having the information accessible prenatally – is often enough to prompt a significant decision around continuing or terminating a pregnancy.

In terms of height, Dr. Banerjee discusses how some parents consider this an issue of social desirability or acceptance, especially as they associate height with gendered expectations for masculinity. Ideas around which existences are socially wanted are transformed into medical concerns, as NIPT reveals SCA conditions and leads to discussions where these phenotypes are framed as medically relevant problems. Genetic counselors also reflected discussions with patients where height became medically relevant and impacted reproductive decision-making:

> Because it's XXY, they have some learning disabilities, they can be tall, they have fertility issues. And then they need testosterone before puberty to help with puberty. […] I had one lady who was really concerned about the height. It wasn't about anything else, just about the specific height. I was like, 'I guess she's probably going to be shorter,' but this lady was like 'how much shorter?' and I'm like 'I can't tell you that.' So I think people will just fixate on different things. (Shannon Interview 2021)

> Height – I've had that experience where some people have focused on the height. Like 'how short is short?' and not really being able to give them that information as detailed as they would want. (Kim Interview 2021)

Genetic counselors Shannon and Kim illustrate instances where patients seem obsessed with height as a factor of their potential child's SCA diagnosis, despite any implications for their health or wellbeing. In many ways, the fixation on height reflects the trends around routinely employing prenatal genetic technologies to identify differences as early as possible, leading to increased medicalization of variations in how individuals exist. Before NIPT illuminated SCAs, parents and providers may not have considered a future child's height as a clinical let alone prenatal concern, unless there were evident structural or developmental issues. But with this technology, height is increasingly medicalized; being too tall or short becomes not only medically abnormal but socially unwanted. Consequently, as NIPT picks up SCAs, and pathologizes related mild and often treatable symptoms, more biophysical traits are made medically relevant, fundamentally changing reproductive decision-making based on conceptions of who represents a desirable existence.

Lastly, and less gendered, is the medicalization around mild behavioral issues that may impact an individual diagnosed with an SCA. Similar to height, behavioral issues may present so slightly (if at all), that it is unclear what their medical implications would be; yet, like infertility, such issues become medicalized much earlier than they otherwise might have been due to being associated with an SCA diagnosis. While behavioral issues may be a concern as a child grows up, there is no certainty around a child experiencing these challenges as a result of their SCA. Further, it is possible they can access extant behavioral or learning supports if and when these issues emerge, without the need for an SCA diagnosis and its associated pathology. However, as both doctors and genetic counselors note, the implication of even the potential for behavioral issues is enough to

suggest to some parents that their child to-be will represent an 'abnormal' existence. Identifying this potential difference, and associating it with an SCA diagnosis, thus frames what may have otherwise been a generally typical life experience as medically concerning and unwanted:

> For XYY terminations - I think they were turned off by the behavioral issues that may occur. (Dr. Leighton Interview 2021)

> "The biggest concern is the increased chance for learning disabilities and behavior problems. That's where people really worry the most." (Mindy Interview 2021)

Considering prenatal SCA concerns around infertility, height, and behavior, we see how biophysical traits that would not have been medicalized in-utero (and may never have been a concern in the individual's life) become relevant for reproductive decision-making. Where these issues may have been absent, mild, or readily treatable at one's relevant life stage, with a prenatal SCA diagnosis, individuals now experience life as though they are 'abnormal.' In other cases, pregnancies may be terminated because such traits are framed as pathological existences with medically relevant consequences for one's to-be child, despite uncertainty around these issues emerging or meaningfully impacting one's life.

NIPT blurs the boundaries between what represents a social concern versus a medical diagnosis necessitating health interventions. Increasing use of the technology to drive agendas around diagnosing genetic differences as early as possible, regardless of health implications, creates an approach to prenatal care and reproductive decision-making that can become focused on ever-constrained social desirability standards, with harmful effects on how we collectively value diverse existences. In the process, reproductive decision-making also becomes more complicated for parents, who are often unsure what to make of SCAs and their potential implications for a child. As more information is available in-utero – and framed as 'medical fact' by way of being part of a clinical testing process – more patient-provider encounters discuss differing biophysical characteristics as concerning enough to impact reproductive decisions.

SCAs and Consequences for Expecting Parents' Reproductive Decision-Making

Access to more prenatal information, particularly information garnered through clinical testing but without clear medical implications, is not always the most beneficial for expecting parents and the providers supporting them. NIPT results around SCAs present numerous challenges in this context, which complicates reproductive decision-making and adds layers of arguably unnecessary stress and complexity to one's pregnancy. Through interviews with patients and their interactions with one another on Reddit prenatal health forums, I show how they experience possible SCA diagnoses and how diagnostic uncertainty can be more deleterious than not having had such information at all. Patients navigate and experience complexities and frustrations around SCA information in three main ways: 1) undertaking substantial independent research to gain a sense of control, 2) pursuing further testing and medical surveillance deemed 'necessary' to disambiguate uncertainty, and 3) feeling stalled given the volume of available genetic data yet without adequate information to be able to interpret the data's significance for their pregnancy. As patients navigate the difficulties of a possible SCA finding, their interactions on online Reddit threads also illustrate them supporting each other as they collectively try to make sense of the new NIPT technology (Barley 1986). Patient experiences show that accessing increased amounts of genetic information

prenatally, especially information without clear medical implications, is not necessarily the ideal prenatal care approach for many families; contrary to physician-researchers' research agendas, having more data, and having it earlier, is not useful for everyone.

I also turn to interviews with genetic counselors, where they echo similar challenges when counseling patients. First, given SCAs often present very mild and wide-ranging phenotypes (if any), with many issues being treatable through extant supports should they become concerns during one's life, counselors report it can be difficult to provide patients clear diagnostic expectations; in turn, parents struggle to interpret this ambiguity in the context of pregnancy decisions. Second, NIPT is a screening tool (not a diagnostic technology), so SCA findings are probabilistic and may also be false positive or inconclusive, potentially complicating patients' reproductive experience further. As a result of these issues, counselors report that many patients regret having learned about a possible SCA diagnosis.

Finally, I discuss how counselors describe patients' decisions to terminate pregnancies with possible SCAs. Given that counselors are typically the main providers supporting patients through reproductive decisions based on prenatal genetic testing results, they illustrate the range of patient experiences and concerns regarding SCAs. And while genetic counselors describe that patients make a range of choices – there is no overwhelming pattern around terminating or continuing pregnancies possibly impacted by an SCA – they illuminate the key apprehensions that expecting parents want to discuss while making related reproductive decisions. In these discussions, gendered pathology and heightened medicalization seep into prenatal decision-making, making it challenging for parents and providers to parse through social biases versus tangible medical concerns. Given the implications that prenatal genetic findings can have for reproductive decision-making and supportive prenatal care, it is critical to reconsider how technologies like NIPT can be more thoughtfully and equitably implemented so that they fundamentally benefit the patient experience, provide meaningful clinical utility, and reinforce inclusive social values rather than detrimental notions of biological determinism and genetic essentialism.

In interviews, many patients did not seem very concerned about SCAs as an NIPT finding. Rather, they were worried about conditions that posed more severe health challenges for their future child, as well as conditions that were rarer yet more severe in implications for disabilities and ongoing medical care. Shanaya, who has been pregnant five times and has one child, recounted her most recent genetic counseling experience: "I remember the amount of time we spent with a genetic counselor talking about (SCAs), and I was so uninterested in that. I wanted to hear about like the Cri-du-chat (Syndrome) and Rett (Syndrome) and, you know, these very, very rare and difficult things. But (the genetic counselor) was like 'Oh, this like X Y confused thing,' and I'm like 'Ugh please, that's not why I'm here with you today!' (Shanaya Interview 2021)." Shanaya pursued a range of prenatal genetic testing, including parental carrier screening, NIPT, and diagnostic CVS. As she notes, sex chromosome variations were not her priority when understanding potential fetal health risks these technologies could reveal. Similar to Shanaya, other patients, particularly those who had received 'normal' NIPT results, did not describe being concerned by SCAs and some did not recall discussing SCAs with their doctors or genetic counselors.

However, this is not to say that SCAs were a non-issue among expecting parents. SCAs posed concerns and complicated reproductive decision-making in cases where NIPT results suggested

possible sex chromosome variations. In other words, when the SCA diagnosis became more of a possibility, parents often began to worry about what this may mean for their future child's health and social identity. Even if expecting parents were not quite sure if or how an SCA may impede their to-be child's life, the uncertainty around diagnostic implications was enough to add substantial anxiety to their pregnancy experience. This played out more clearly in patient interactions on online prenatal health forums, where they readily describe their personal experiences and often solicit feedback from others. In the following examples, we see that even though SCAs often do not materialize in fetuses or future children, the suggestion of potential 'abnormality' nonetheless causes significant stress.

A main way patients grappled with uncertain SCA findings was via substantial independent research efforts to understand SCAs, NIPT related capacities, and determine how concerned they needed to be. They often used this information to continue engaging with providers, challenging initial assessments or asking for further clarification. One of the most extensive and engaged discussions illustrating expecting parents' frustrations and anxieties around SCA findings took place in the Reddit "Clinical Genetics" forum (also referred to as a 'sub-Reddit,'), which is described as a space for discussions about "Genetics and Human Well-Being." In this thread, which received 30 individual replies, an expecting father detailed the experience he and his pregnant wife had with NIPT and a possible finding of Turner Syndrome (Monosomy X). The expecting father seems concerned about NIPT's accuracy and capacity as a cell-free DNA (cfDNA) screening tool, pushing back on the genetic counselor's assessment of their to-be child's risk for Turner Syndrome. Throughout, he tries to not only situate his to-be child's risk more accurately and personally, but he pursues a substantial amount of genetics research in order to temper his worries for his to-be daughter. This effort is not inconsequential, as it illustrates how patients must find ways to cope with prenatal genetic information that is probabilistic and unclear in its medical implications yet amounts to challenging reproductive decision-making.

The father starts his post stating, "After my wife's NIPT (Harmony, *genetic testing company*) returned positive for Monosomy X, we dug into the research extensively. We disagree with our Genetic Counselor's assessment — ballpark 40% chance baby has Turner Syndrome. […]. Conditional on relevant factors from the Turner Syndrome literature, we believe our specific probability is far lower." Based on his research, he further problematizes that NIPT is a screening tool not well-suited at identifying whether an SCA is indeed within the fetus (or if the variation is derived from another source): "As we understand it, cfDNA screens (*cell-free DNA screening technologies, e.g. NIPT*) are extremely effective at determining that a "missing X" exists somewhere in the maternal blood sample. […] The possibility of a false positive is almost entirely due to the possibility that the dna with the missing X might originate from sources other than the fetus: from the placenta, a vanished twin, or the mother." It is evident that this father has taken notable steps to understand NIPT, illustrating the significant determination some patients go through to better understand their prenatal testing results and novel technologies in the context of consequential reproductive decisions. This poster has conducted extensive research on his own and tries to triangulate this information with the specifics of their pregnancy (e.g., lack of ultrasound findings) and their family history: "None of the classic markers of Monosomy X are present. […] A few weeks later we spoke with the Chief Geneticist. He agreed with this analysis, citing the normal (nuchal translucency) and clear ultrasounds so far as the encouraging signals he

was seeing. Since baby was high risk for Turner, they did a level 2 fetal anatomy scan at 20 weeks, came back clear." (Reddit Patient Forum 2021, *clarification added*).

At this point, the father delves into nuanced risk statistics based on his independent research, which is a common approach among concerned parents hoping to mitigate their anxiety around having a child affected with an SCA. Moreover, as described, the father escalates his concerns to the Chief Geneticist, illuminating how expert opinions matter during patients' reproductive decision-making; these perspectives carry power as they represent scientific fact and can suggest to parents that their concerns are valid and worth acting upon (or that their anxieties may be quelled, as in this case). "I said to the geneticist, 'From what I can tell, these indicators in combination ought to put her real risk at under 5%' (Honestly I put the number at <1% as the best guess based on available information, but I was more interested in hearing whether an accomplished authority on the subject thought I was in the neighborhood than quibbling over a reasonable cutoff number). He said while available research leaves him unable to put a number on it, 'You may well be right on that. I don't think it's a false hope.'" Ultimately, the father updated his post: "Baby recently arrived. Healthy as can be. And a week later, we received word on the karyotype from Genetics: Our daughter has both X chromosomes, and is genetically normal." He goes onto say that his wife is considering getting genetically tested, as she wonders if the "missing X" could have come from her. For this poster, it seemed like researching NIPT as a tool and diving into risk statistics around Turner Syndrome (as well as pursuing further testing and medical procedures, which I discuss momentarily) made all the difference in their ability to understand their to-be child's health and make a reproductive choice aligned with their values. (Reddit Patient Forum 2021).

This relief did not come without much exertion and self-advocacy on the patients' part; the substantial research patients undertake not only requires notable effort but a certain level of health literacy, which not all patients are privileged to have. Many commenters also critiqued the father's fixation on unraveling the Turner Syndrome risk statistics in this way, encouraging him rather to consider the "bigger picture" around what this potential SCA could mean for his to-be child and whether that aligns with his and his wife's reproductive values. One responder told the father to "go with his gut" on whether he and his wife would want to raise a baby with possible Turner Syndrome, regardless of the numbers around the child's risk of having the condition. Commenters stated that this father was "too caught up on the numbers," and labelled his approach as a "coping strategy" common to many parents anxious about NIPT SCA results that are probabilistic and ambiguous. Offering support, several commenters also reminded the father that NIPT is a screening tool with false positives. For the father, however, his commitment to unpacking specific implications of the NIPT Turner Syndrome finding, effort toward disentangling the genetic counselor's initial 40% risk estimate, and consistent advocacy around getting further testing and more 'authoritative' genetic perspectives seemed like a 'win' against the murkiness of NIPT and SCAs. Indeed, he later responds to another poster in a similar situation to express his happiness that their risk estimation was also inaccurate and that their child was born without an SCA despite NIPT suggesting otherwise. (Reddit Patient Forum 2021).

SCA findings can lead to prolonged periods of stress and concern during pregnancy; beyond the additional worry expecting parents face regarding their fetus' health (and research efforts often taken to mitigate this), SCA findings on NIPT can also lead to further medical interventions for a pregnant person. After NIPT identified a possible SCA, it was common for patients to undergo

more testing to verify if the fetus indeed presented with an SCA, increasing the scope of medicalization into their pregnancies. For example, a Reddit poster shares: "I was NIPT positive for turners syndrome. My MFM pushed me to wait for amnio because I had no fetal markers on US. On amnio they found normal chromosomes. They felt it was likely the placenta was mosaic for turners. I have a typically developing 2.5 year old now (Reddit Patient Forum 2021)." While diagnostic testing was useful for this patient, we see how NIPT, in providing results for Turner Syndrome that were probabilistic and non-specific as to whether the 'missing X' was indeed within the fetus, led to further medicalization of her pregnancy. Pursuing additional testing and medical procedures to resolve uncertain SCA findings is consequential for patients; it adds medical interventions, associated financial costs, stress during waiting periods, and emotional burdens of having to process one's pregnancy as possibly abnormal. In another thread, a poster discusses how her NIPT test was "inconclusive" for fetal sex, describing a series of possible further prenatal testing interventions and her concerns that the to-be child would be 'abnormal':

> "I met with a genetic counselor which basically said everything is probably fine, the lab was just unable to confirm with confidence what the sex is, but there is a chance there is a sex chromosome disorder. (*My genetic counselor*) suggests waiting until my 20 week scan where they should be able to visually identify the sex and also if anything else looks off. If anything looks abnormal at 20 weeks then I can opt for an amniocentesis test to confirm the sex and any chromosome abnormalities. From what I googled it seems like the inconclusive results are more common with higher bmi's (*body mass indices)* so I am hoping that's all it is (I have a very high BMI, *body mass index,* of 58). Has anyone else received an inconclusive sex result and how did everything turn out in the end? I am trying not to freak out because my 20 week scan isn't until December 16th. (Reddit Patient Forum 2021, *clarification added*)."

Having access to SCA information prenatally, paired with the uncertainty of its diagnostic implications, can be a source of consistent anxiety during one's pregnancy – this is compounded when additional testing and medicalization are introduced. Despite her counselor's reassurance, this pregnant person remained concerned about an SCA if only because of the potential for a diagnosis (irrespective of actual health implications for her future child) and turned to further medicalization as a way to resolve uncertainty. She shares the additional prenatal screens and diagnostic tests she may pursue, while saying she is "trying not to freak out" during the waiting period. Similarly, other patients, including those who were interviewed, reported the waiting periods in between prenatal tests to be incredibly challenging, adding persistent worry to their pregnancy experience. In this case, even though the patient received inconclusive (not 'abnormal') results, they continued to focus on the possibility that the fetus might have structural issues or genitalia that "looks off," and made plans for further testing in the event of their fetus' sex being atypical. Here, we see how expecting parents turn to additional medicalization to cope with SCA findings and uncertain diagnostic implications, indicative of the stress that they endure as they try to reassure themselves of a healthy pregnancy – an idea that, for this patient, has been tarnished by even the possibility of sex chromosome variation.[44]

[44] Like other patients sharing their experiences on Reddit as well as patients interviewed, this poster also describes Googling what a possible SCA finding might mean. In order to situate the "inconclusive" fetal sex finding for her specific reproductive experience, this patient turns to research suggesting that her body mass index (BMI) may play a role in her results (rather than an actual SCA diagnosis for the to-be child). Here, we see another example of patients

A third way SCA findings on NIPT can complicate patients' reproductive experiences relates to the volume of genetic data made available to them without adequate and appropriate resources to be able to make sense of such data. In other words, patients have too much data with too little information to interpret it. Often, this issue is reflected in how patients struggle with decisions to terminate or continue pregnancies with SCA findings on NIPT. Not only are SCA findings distressing because of the ambiguity around whether and how severely the fetus might be affected, but expecting parents must try to envision the life their child may have with a condition that is so mild or vague that it could never present or be easily treated during their life. In these moments, parents can face challenges around understanding SCAs as a medical condition, with diagnostic implications, and this added stress shapes how they approach complex reproductive decisions. Below, patient Clarissa describes how she supported her niece through a challenging and uncertain SCA finding on NIPT. Clarissa is a fervent disability justice advocate, disability community and non-profit leader, and mother to three children, one of who has a chromosomal condition. She explains that her niece did not know that NIPT might reveal sex chromosome variations because she like so many others understood the screening to be a "gender test" as it is popularly positioned. As Clarissa's niece turned to her for support around how to interpret a possible Turner Syndrome finding, worried yet hesitant to end her pregnancy, Clarissa became a central resource:

> I just had a niece who took the NIPS (NIPT). And it came back with high probability for Turner Syndrome. She's in her 20s. When we actually ran her positive, predictive value in the Parental Quality Foundation Tool *it was like an 84% chance for a false positive*. So, I told her, 'I know it's concerning when you get information like this. But really this is what life of Turner syndrome looks like,' and *I gave her some resources that our organization had created* […]. *She felt kind of deceived*. Because you take this test, and you think you're going to get some sort of definitive results. […] *But to know how inaccurate they can be for the microdeletions and sex chromosome conditions, it's very concerning for patients.* […] *That causes a lot of stress and anxiety in a lot of ways*. […] People are getting all of this genetic information that they aren't prepared for. *What (my niece) told me is that she thought it was just going to be a gender test and now she has this information about Turner Syndrome that she does not know what to do with.* She was not given information about Turner's Syndrome (by her providers) […]. (Clarissa Interview 2021, *emphasis added*)

Clarissa's niece pursued NIPT without being thoroughly counseled on the types of information about genetic variations that the test could identify; subsequently, she was faced genetic data about an SCA that she did not know how to understand. Clarissa describes her niece feeling "unprepared" and "deceived" to learn that not only could a "gender test" reveal a possible genetic condition, but that this sex chromosome variation may not actually be present or meaningfully impact their child's life. In fact, according to genetic counselors, many patients are unprepared for the possibility that NIPT may reveal an SCA as a possible diagnosis, rather than simply a "gender test" that tells them whether they are having a boy or a girl. Despite being caught off-guard, patients must process this uncertain and highly probabilistic information in light of consequential reproductive decisions; fortunately Clarissa's niece had access to robust resources and support via

undertaking independent research efforts to manage SCA findings, uncertain diagnostic implications, and mitigate stress in their pregnancies.

Clarissa's disability justice non-profit and expertise. However, for others without such networks and access to relevant providers and information, wading through reproductive choices in the face of significant ambiguity can be a far more daunting. As Clarissa notes, her niece did not receive disability education resources from her providers, but instead had to rely on Clarissa for these supports to better understand how her future child's health might be impacted and make an informed decision around her pregnancy (she chose to continue). While many physician-researchers continue to push for more genetic information to be made available prenatally, it is worth questioning whether this 'more is more' approach is helpful to patients who must contend with the data in the context of reproductive choices.

These patient experiences urge us to reconsider the value of SCA findings for pregnant patients. Is information highlighting a genetic difference valuable simply for the sake of having such knowledge? In the accounts above, patients describe that having information about possible SCAs can be more stressful than beneficial, especially when the health implications of these variations remain uncertain. Expecting parents may turn to extensive and burdensome 'research rabbit holes,' pursue more medicalization and testing, or feel stalled given the amount of genetic data they need to contend with in light of reproductive decisions. As such, having access to sex chromosome variation findings, even without health implications, can be difficult and complicated. Instead, an alternative approach to delineating fetal health, perhaps more focused on clinical ultrasound markers or more severe conditions with defined health implications, could be more medically useful for prenatal care and reproductive decision-making.

Before turning to genetic counselors, I want to highlight how patients are important resources for one another on online prenatal health forums, as this is an important way that they navigate the challenges of SCA findings described above. These forums are a crucial avenue of compassion, education, advocacy, and reassurance among parents going through challenges in their prenatal genetic testing journeys. And, for many who do not have access to the medical experts and resources they need, these forums become invaluable sites for understanding genetic differences and disabilities. There were several posters supporting other individuals who shared anxieties and concerns about possible SCA diagnoses. As patients described their frustrations around how to interpret SCAs as a medical condition, and raised questions around what the implications may be for their child's future health and social identity, commenters often provided nuanced and nurturing perspectives. Recall the Reddit poster who shared her experience about inconclusive fetal sex results on NIPT as she wondered if her body mass index might have contributed to this finding.[45] There were five people who responded, each providing support and reassurance while also raising critical points about sex and gender. One response received several 'upvotes' (similar to 'likes' indicating agreement) from other online community members:

> "*Gender is a social construct, much different than sex determined by genetics* […]. The reason it is checked in genetic screening is because it is complex and *requires a lot of learning and understanding and accepting*. *Many children born with (SCAs) do not look any different at birth.* Putting myself in your shoes, *I can imagine the frustration and*

[45] This pregnant person shared her experience on a popular Reddit forum called "CautiousBB" (colloquial for 'Cautious Baby'), which is described as a discussion space "For cautious pregnant people on this great, perilous journey to parenthood."

> *anxiety in just being given an inconclusive result*. […] Since it sounds like the genetic counselor wasn't convinced there is an issue, and it can be more common than people realize to have an inconclusive result from these kinds of tests, and a variety of biological and environmental factors can affect the results, I would remain hopeful that everything will turn out okay in the end. […]. (Reddit Patient Forum 2021, *emphasis added*)."

This responder provides not only support and nurturing words, but shares a thoughtful perspective on sex, as embodied biological features, as distinct from gender being a "social construct." They describe SCAs as requiring "learning and understanding and accepting," positioning this possible diagnosis as more of an adjustment of one's social expectations rather than a medical concern. Finally, the responder provides comradery to this worried pregnant person, empathizing with her feelings of "frustration and anxiety" given the inconclusive NIPT result. They offer reassurance, citing that inconclusive results are common with NIPT as a way of perhaps indicating to the pregnant person that they are not alone, and encourage the pregnant person to stay hopeful.

The response received positive attention from other forum community members, with one individual directly replying: "Yes! People, stop conflating sex and gender. Someone asked me yesterday if I know my babies gender yet and I said 'of course not and I probably won't for another 3 years? At which point they can be whatever gender they want *to* be. Her sex is female though (and until she becomes self aware or tells me otherwise, that's how I will refer to her).'" In this way, patient communities not only offer meaningful support, valuable information, and compassion to one another, but also provide critical and nuanced perspectives on sex and gender in the context of medicalized SCA diagnoses – discourses that are less salient in clinics and genomics conferences. (Reddit Patient Forum 2021)

Moving forward, I turn to how prenatal genetic counselors help parents through NIPT findings of sex chromosome variations and the convoluted reproductive decision-making that can follow. The data comes from interviews with genetic counselors at a range of practice types in California, from large private and academic hospitals to public hospitals and non-profit clinics for low-income communities.[46] Across these practices, genetic counselors are the key resources for patients, and their perspectives on how patient discussions unfold capture relevant points of apprehension and uncertainty in patients' experiences. These counselors report that one of the main reasons patients struggle with SCA findings is due to the phenotypical variability in how these conditions present; reproductive decision-making can be difficult if patients cannot ascertain whether or how severely their future child might be affected. Similarly, counselors note that NIPT can often return inconclusive or false positive results for SCAs, further complicating patients understanding of their pregnancy and subsequent decision-making. As such, counselors described patients often regretted having information about SCAs; indeed, more information is not always more useful. Accordingly, many counselors shared in interviews and conference presentations (as discussed earlier) that NIPT should not be used to routinely screen for SCAs. They explain that the uncertainty around the information drawn is not appropriate or useful for all parents, especially when it does not have clinical bearings for the providers supporting them.

[46] The counselors interviewed for this research practiced at various clinical settings in California: large private hospitals, general public hospitals, non-profit clinics for low-income communities, research-focused clinical settings, fertility and prenatal specialty clinics, genomics company affiliated counseling centers, and counseling centers affiliated with private physician practices.

Similar to expecting parents' personal accounts, genetic counselors describe that their patients struggle with making reproductive decisions based on SCA findings because of the wide variability in how these conditions may present. Counselors note that milder conditions like SCAs can often be harder to interpret in the context of one's family expectations, views on disability, or medical needs for a future child because it is unclear to what degree an individual might be affected. Mindy, a genetic counselor at a large public hospital, explains: "People don't seem to usually need help that much (around termination decisions). […] But sex chromosome conditions are the ones that people struggle with the most because they're variable, because they're more mild, than Down Syndrome is. People do seem to have a harder time with (SCAs) (Mindy Interview 2021)." Below, genetic counselor Hannah, who works at a general hospital, explains SCA variability further:

> "Once you get a positive result for Turner Syndrome it is super hard because it is 'this baby is not viable,' which is very possible. Or 'you haven't had an ultrasound since six weeks, we need to get you in for an ultrasound.' Or, maybe the baby could look totally normal on ultrasound, and it could be a false positive. It could be mosaic and end up fine and they would never know otherwise. There's a big, big range of possibilities from fetal death to like pretty normal person. So, for (SCAs), when I have the positive NIPTs, we have multiple things we have to figure out. Like, these are the possible etiologies of this positive result, it's either coming from (the pregnant person), from the placenta, or from the placenta and the fetus. We have to figure out where it's coming from. […] (Hannah Interview 2021)."

Hannah illustrates a wide range of outcomes associated with a possible Turner Syndrome finding, describing how this can make counseling patients challenging. There is not only the question of whether the SCA is indeed affecting the fetus, but also the uncertainties around how severely it may present if it does materialize phenotypically. Similar to other genetic counselors, Hannah's approach relies on the pregnant person undergoing additional medical interventions and surveillance to delineate where the chromosomal variation is coming from. Once such uncertainties have been addressed (*if* a patient wants to pursue, or can afford, further testing), a patient might then be able to move forward with reproductive decisions based on their priorities and values. In cases where further testing is not possible or desired, uncertainty continues to loom over the pregnancy. As parents struggle to make sense of the variability and ambiguity of SCA findings in the context of their reproductive journeys and family visions, this is reflected in how they may approach decisions around termination.

Genetic counselors also emphasize that the ambiguity around how NIPT presents possible SCA findings – being probabilistic, possibly false positive, or inconclusive – leads to substantial stress for patients during pregnancy. There are also instances where sex chromosome findings on NIPT do not correspond with how providers determine sex per ultrasound, which can confuse or worry patients. Given that many patients do receive NIPT results that are inaccurate or inconclusive regarding their fetus' potential SCA, this is a significant source of complexity and stress during one's pregnancy. Below, Kim, who works as a genetic counselor for a private genomics testing company, describes a patient with SCA findings who felt unprepared to deal with such ambiguity but had to make consequential reproductive decisions nonetheless:

> "I had (a) situation where the NIPT was showing a male gender, but they looked like a female on the ultrasound. So she did the amniocentesis. *She was very close to the 24-week mark, so she had to make a decision. […]* She did the NIPT like three times, and the first time came back uninformative sex and then the other two times Y chromosome was detected. […] But then the final result (amniocentesis) didn't show any cells with Y chromosome. *And so, you know that already would have been really confusing to her.* […] *I don't know if you can really relay to someone that even if you do a pretest counseling that you might get some results that's not very informative or that's going to confuse you even more.* (Kim Interview 2021, *emphasis added*)."

Kim explains that it is challenging to counsel patients because NIPT can return a range of complicated and confusing results around SCAs. For her patient, NIPT provided three conflicting findings around a possible SCA; however, the diagnostic amniocentesis showed the fetus to have typical X chromosomes associated with female sex. Even though the patient pursued further testing via amniocentesis to clarify the SCA results, they terminated the pregnancy before these results were ready, as they were approaching California's 24-week termination timeframe and the ambiguity was enough to make them consider their pregnancy and fetus to be unhealthy or abnormal. Ambiguous SCA results with unclear medical implications not only add stress to a pregnancy but can have tangible consequences when expecting parents choose to end a desired pregnancy based on a genetic difference that, when presented as a part of routine prenatal clinical screening, implies abnormality. Ultimately, Kim reflected on how she was not sure what she could have done differently, questioning whether she could have appropriately prepared her patient to deal with this uncertainty so that the pregnancy, which was eventually found to not have an SCA, may not have ended in termination. In such cases, we see the importance of implementing prenatal genetic testing tools critically, with caution; the information that tools like NIPT reveals can have substantial consequences on patients' lives and reproductive journeys, and it is worth reconsidering what types of conditions to routinely screen for, possibly omitting those SCA conditions that the technology is not yet well-suited to detect reliably.

As a result of these challenges around NIPT and SCAs, counselors described that having genetic data about SCAs, especially data with ambiguous implications, is not always beneficial for patients, some of whom regret having learned about the condition at all. Where a sex chromosome variation may have never been consequential for one's future child and family, having knowledge about the genetic difference prenatally means that parents need to process their pregnancy and family vision in the context of an implied abnormality. Many counselors recounted patient encounters where simply having access to prenatal information about a possible SCA, despite the condition not being diagnostically confirmed or presenting health challenges, led to anxiety that negatively shaped pregnancy experiences. In these instances, having more information prenatally is not necessarily the most helpful approach for expecting parents. Emma, a genetic counselor at a large university medical center, describes this anxiety and apprehension she helps patients process:

> "We have had some people who have sex chromosome aneuploidy results, and that has ended up causing them some anxiety. Some people find out more about the particular condition, and then make their decision about whether they want to do more testing. But some people just […] accept that (the fetal sex) is definitely not in the category that (they) are looking for in prenatal screening. *So they kind of have a situation where there is a bit*

> *of regret about finding out this information and then having to incorporate it into their pregnancy. […] That's one of the reasons that sex chromosome aneuploidies do cause more anxiety, just because you know this information*. Maybe you don't feel like you would change anything about a pregnancy based on that, but you still now know this information (Emma Interview 2021, *emphasis added*)."

As Emma describes, patients can regret learning about SCAs, as the information alone (regardless of health implications or reproductive priorities) is enough to cause anxiety and recast their pregnancy as possibly unhealthy or abnormal. Even if an expecting parent may not want to terminate their pregnancy based on an SCA finding, they can experience heightened stress as they contextualize their pregnancy and future child's identity as potentially pathological. In other instances, having access to uncertain or unclear information about SCAs can be more challenging for patients and lead to terminations. Genetic counselors describe these cases as being particularly difficult to counsel patients on, as they are unable to provide adequate information about if or how the future child might be affected, leaving parents to contend with uncertainty. As such, genetic counselors suggest that routinely screening for SCAs prenatally does not always provide patients clinical benefit and can hinder informed reproductive decision-making.

Even though many pregnancies with SCAs reach full gestation and can be considered overall healthy, an SCA finding often prompts counseling sessions around whether a to-be child will be 'normal' and the reproductive choices a parent in California may want to consider (where terminations are legal until the 24th week of pregnancy, and at any time to save a pregnant person's life). Most often, genetic counselors are the key providers supporting patients as they process decisions to continue or terminate a pregnancy and are specifically trained to provide balanced non-directive guidance. Importantly, because reproductive choices are so personal, counselors emphasize that expecting parents make a range of decisions when faced with an SCA. That is, there is no discernible pattern around *what* parents choose; while some continue their pregnancies, others terminate, illustrating how even the suggestion of difference implying abnormality can be consequential. However, there are identifiable themes around *why* they pursue their decisions. Understanding what leads patients to their reproductive choices not only reveals the implications of framing a genetic difference as a pathological medical diagnosis, but also illuminates how social assumptions about appropriate or desirable existence are transformed into clinical 'facts.'

Counselors repeatedly pointed to three main concerns around SCAs that led patients to termination: 1) structural issues within the fetus that would tangibly threaten the health of the pregnancy or necessitate involved medical interventions for the future child, 2) anxieties around gender that did not align with how parents envisioned their family or expectations for their future child to represent a 'normal' social existence, and, 3) as somewhat related to gendered anxieties, parents fixated on ideas that their future child would be predisposed to deviance and social rejection, dredging up 'criminal chromosome' stigma despite such research being long debunked. I explore each of these below.[47]

[47] Other points counselors raised around parents' reproductive choices related to concerns around height, infertility, and behavioral or intellectual issues, which have been discussed earlier in the chapter.

First, providers reiterate that patients are typically most concerned when ultrasound findings indicate that an SCA poses accompanying structural issues for the fetus that could jeopardize the health of the pregnancy and require involved medical interventions for the to-be child. Some specific concerns that emerge relate to heart defects, issues with gonad development that are associated with malignant cancers, cystic hygromas,[48] and hydrops fetalis.[49] As such, while doctors and genetic counselors typically present SCAs as relatively mild conditions with little to no health impacts on the to-be child, in cases where anatomical development is affected, providers are more concerned around how to manage these pregnancies and patients often do terminate pregnancies to avoid miscarriage, stillbirths, or challenging medical interventions and life experiences for the to-be child. As the genetic counselor below describes, these concerns often emerge alongside a possible Turner Syndrome (Monosomy X) diagnosis:

> I see termination mostly with Turner Syndrome diagnosis and that's just from the standpoint of babies with Monosomy X often have severe abnormalities. They usually have cystic hygromas or they have hydrops, fetal pleural effusions, so a lot of those babies just in general don't` make it. […] And so […] for Monosomy X, where we're seeing those abnormalities, a lot of people do interrupt. Not because of the Turner Syndrome diagnosis itself, because a lot of people where there's not those findings do continue (pregnancies) with Turner Syndrome. But unfortunately most of those babies do have those types of ultrasound findings. (Rosemary Interview 2021).

Rosemary emphasizes that expecting parents may not terminate their pregnancies given a Turner Syndrome diagnosis itself; it is when the Turner Syndrome finding is paired with concerning ultrasound findings regarding the fetus' structural development that many patients terminate. Several counselors and physicians echoed this perspective, explaining that Turner Syndrome in particular seemed like an exception to the relative mildness of other SCAs.

Second, genetic counselors also note that non-binary presenting genitalia and secondary sex characteristics can be a concern for expecting parents. As discussed, imaging and classifying genitalia per binary sex is a priority among reproductive physicians, who associate 'typical' binary appearing genitalia as an indicator of a 'normal' healthy pregnancy. This focus on genitalia to determine normalcy translates to expecting parents. Often, expecting parents associate non-binary genitalia or genitalia that does not correspond to one's sex chromosomes with expectations for their future child's gender identity, parenting approaches, and social expectations. A misalignment between genitalia and sex chromosomes, especially in the presence of a potential SCA, can suggest to parents that their child will have a 'deviant' gendered existence, filled with heightened stigma and challenges, despite that non-cisgendered individuals lead fulfilling lives in inclusive and equitable social environments. Similarly, parents were concerned about secondary sex characteristics and gonads that appear non-binary or 'discordant' with their fetus' other sexed

[48] A cystic hygroma "appears as a sac-like structure" "that most commonly occurs in the head and neck area." Research shows that many fetuses with hygromas do not make it past 26 weeks of gestation, but for those that do there is a 67% "chance of ultimate survival" (Anderson and Kennedy 1992).

[49] Fetuses with hydrops present with "extensive swelling" due to a large amount of fluid accumulated in their tissues and organs. Hydrops carries a particularly high fetal mortality rate, with 60% to 90% of these pregnancies ending in fetal demise ("Hydrops" n.d.).

features and chromosomes: “People have been concerned about possible gynecomastia (enlarged breast tissue in males), as well as possible smaller penis. (Rosemary Interview 2021).” Embodied sexed features, and related assumptions around sex and gender, play a key role in how patients establish normalcy for their pregnancies and future children, informing reproductive decisions and medical interventions accordingly.

Genetic counselors describe that these issues around sex and gender ‘discordancy’ can be challenging for some patients largely because they are unfamiliar with concepts around sex and gender and how individuals may embody diverse existences. “(Things like ambiguous genitalia are) harder for patients because it's often something that they have never dealt with themselves and don't know how that functions in the world and what that life would look like for someone who's dealing with that. (Hannah Interview 2021).” Counselors note that some patients in these situations opt for termination, while others seek additional support and resources for better understanding. Here, we see the importance of wider disability and social inclusion in understanding to what extent a genetic difference needs to be medicalized and how it may inform reproductive choices, as expecting parents’ exposure to those with diverse sex and gender identities can shape how they grapple with SCAs during their pregnancies.

It is worth noting that as patients and counselors discuss concerns around ‘discordant’ sex and gender, gender is once again pathologized in these consultations (as it was between patients and doctors in clinic). Below, genetic counselor Mindy details her approach to counseling patients whose fetus presents an SCA and non-binary genitalia; while she focuses on reassuring patients, social expectations around sex, gender, and sexuality are continually pathologized in the process, reinforcing harmful stigma:

> “*I will have people ask, ‘does this mean my baby is a hermaphrodite?’ and then I'll say, ‘The term is intersex these days*. Most people with this sex chromosome difference are not intersex,’ and then I would say, ‘Your baby has XYY chromosomes, that is male. *And so your child's gender identity is not any less likely to be male than any other boy.*’ Or ‘your son does have an extra X chromosome, men and boys with Klinefelter’s syndrome *do have some traits that may be considered more feminine, but they typically still have a male gender identity.* And many people with Klinefelter’s syndrome *are attracted to women, are heterosexual, and marry women, but for any child there's a chance that they may not be heterosexual as well*.’ […] Enough people have (asked) that I will try to say, once it's a diagnostic testing result, ‘just so that we're clear. This means that the baby is female, and has (SCA), but she's still female.’ (Mindy Interview 2021, *emphasis added*).”

Mindy, like most counselors, is seeking to be supportive, empathetic, and balanced in supporting her patients. She repeatedly clarifies appropriate terminology and tries to reflect a wider and more inclusive approach to sex classification. Still, in doing so, her counseling around sex chromosomes, genitalia, and gender conforms ‘normalcy’ into a male/female sex binary that corresponds with a cisgender identity. Although she may be taking this approach to comfort patients, she inadvertently perpetuates the notion that individuals whose sexed biological features and gender identities do not align are abnormal to some extent. Further, she also presents stereotypical assumptions regarding certain traits being considered more feminine or masculine, as well as setting heterosexuality as a normative expectation. There is an implication that parents ought to feel

reassured that their future child will embody and reflect normative sexed features and cisgendered heterosexual identities, with those outside these bounds representing abnormal or unwanted existences. As such, in trying to counsel and help patients through their anxieties around SCAs, providers and patients continue to pathologize diagnoses based on sex and gender assumptions that are fundamentally social. As a result, the distinction between 'normal' and 'abnormal' is further defined, with less social acceptance for those whose sexed features differ from their gendered and sexual identities, and greater medicalization of these existences as pathological.

Third, genetic counselors describe that expecting parents often raise concerns around gendered expectations related to deviancy and delinquency as genetically essentialized traits. Although research on the "criminal chromosome" has been debunked, these medical myths are perpetuated when parents navigate implications of a possible SCA. Such myths reflect gendered biases, as they make harmful assumptions about what it means to be a "Super Male" with an extra Y chromosome, attaching deviant maleness to traits of aggression, social incapacity, and a lack of empathy. Genetic counselors describe how these gendered assumptions and medical myths continue to permeate as parents learn about SCAs, at times with significant consequences around termination. Below, Rosemary discusses her experiences counseling patients faced with an XYY sex chromosome variation (Jacobs Syndrome):

> There are some people that are very scared, particularly, I've noticed for XYY. I think because *there's still some of that old literature that's out there that associated with criminal behavior*. There were some studies back in the 70s that linked men that had XYY to having an increased risk for criminal behavior. I think some of that literature is still out there that when people search. *They get a little bit scared when they've done their own research*. We provide them with as much information as we as we can and tell them to steer clear of this (research). *But we had one patient recently, she had XYY, did an amnio and decided to interrupt the pregnancy. She was scared about that possibility (of criminal behavior), even though we did as much as we could to disabuse her of that.* It was just unfortunate, because all of that research has been completely debunked […] but I guess people can still find it. (Rosemary Interview 2021, *emphasis added*)

Despite counselors emphasizing that these medical myths were indeed myths, and had been scientifically falsified, the association between criminality and an additional Y chromosome held power in pathologizing these SCAs within patients minds. That these debunked ideas continue to circulate once they are established is consequential; as Rosemary describes, her patient terminated her pregnancy based on fears that her to-be child would be genetically predisposed to criminality.

As SCAs are pathologized into medical diagnoses based on normative social assumptions about sex, gender, sexuality and related tropes around masculinity and femininity, the boundaries around which existences we consider normal and desirable versus those seen as abnormal and perverse are continually sharpened. In the process, social biases are conflated with medical diagnoses, lending scientific authority to 'facts' that are in themselves built on changing ideas about identities and inclusivity. SCAs thus come to be seen as objectively pathological medical diagnoses, when indeed the very assumptions underlying their creation as a diagnostic category are socially subjective and mutable. The consequences for reinforcing a diagnostic category in this way are meaningful. In the case of SCAs we see parents struggling to navigate uncertainty, heightened

medicalization of pregnancies and infants, resulting reproductive decisions to continue or terminate pregnancies, and providers who are unsure of how to guide parents appropriately. Taken together, the rift between normal and abnormal becomes gradually more rigid, with lasting ramifications for our values around inclusivity and tolerance, and the continued marginalization of those who represent disabled or different existences in this increasingly narrow context.

To be sure, it is not that any particular group is single-handedly leading the pathologizing SCAs or doing so with the goal of marginalizing those with diverse sex and gender identities. Rather, we see how these processes play out collectively, during interactions, at various medicalized venues, and among both patient and provider groups. In many ways, these individuals are often well-intentioned, with genetic counselors and doctors attempting to support their patients in clinics, patients leaning on one another in online health forums, and physician-researchers fervently pushing forward research agendas they believe will lead to worthwhile treatments for those diagnosed with SCAs (and, indeed, many of these early interventions do make a difference in the lives of infants with SCAs and their families). However, their discourses, discussions, and medicalized interactions continue to reinforce one another in establishing the notion that binary sex and cisgendered existences are a medical norm and therefore the idealized desirable social existence. The assumption becomes that one's sexed features – e.g., genitalia, gonads, secondary sex characteristics, sex chromosomes – must not only align with one another to classify an individual per male/female binary sex, but that this classification should then conform to one's lived gender identity as a man/woman where certain traits are typified as masculine and others as feminine to mark an appropriate gendered existence. That these processes unravel in scientific and medical spaces implies objectivity to their conclusions, even as scientific and sociological research has long established the substantial variation in how sexed features may present and exist in 'discordancy' from one another and eventual gender presentations. As such, even though our contemporary sociocultural attitudes around sex and gender seem to become more inclusive in some ways, in other spaces (e.g., medico-scientific spaces imbued with notable power, authority, and social legitimacy) we may be seeing a gradual return to reductive and harmful notions.

Novel genetic technologies like NIPT play a critical role in how these collective interactions among patients and providers unfold. The process of pathologizing sex and gender among patients and providers is centered around NIPT as a new tool, bringing with it new opportunities, in prenatal clinical care (Barley 1896). NIPT not only enables more capacities (e.g., routinely and relatively easily screening for chromosomal variations earlier in pregnancy), but it also engenders new problems as the technology is implemented widely and perhaps too rapidly without considering the realized clinical value of each finding and the resulting social and medical consequences for prenatal care and patients' reproductive experiences. Technologies like NIPT, with their effects on how diagnostic categories like SCAs are medicalized based on social biases, raise questions around what information is tangibly useful and beneficial for pregnant patients and their providers. Just because information about genetic differences can be accessed prenatally, does it mean it's worthwhile to do so? Moreover, simply because a genetic difference exists (as it does across all individuals), does this difference need to be medicalized and pathologized, especially when it may not substantially impact one's life experience? While some may value information for its own sake, for others, having this knowledge can make reproductive experiences quite complicated as parents struggle to interpret SCA findings in the context of their family visions and providers struggle to adequately support patients. Given that prenatal genetic information, particularly about

mild and variable SCAs, can be more deleterious and stress-inducing than beneficial, it is worth being more thoughtful about how new prenatal technologies are utilized in light of implications for individual reproductive experiences, clinical care, and broader societal consequences.

Final Thoughts on Reinforcing Sex Chromosome Aneuploidies as a Prenatal Diagnosis

Sex and gender have always played an important role in our sense of identity, but more importantly in our relationships with and expectations of one another. Pregnancy is a salient moment wherein sex and gender become pronounced. Providers use markers of fetal sex to establish the normalcy of a pregnancy, while expecting parents seek to learn 'what' they are having – indeed, the moment where a doctor declares whether a fetus or newborn appears to be a boy or girl has long captured popular imagination about reproduction and childbirth. For expecting parents, much of this excitement revolves around understanding who their future child will be and how they envision parenting this child based on its assumed gender. In this way, sex and gender are meaningful foundations of how we develop our identities in relation to those around us. Being able to present or 'do' one's gender 'appropriately,' in accordance with learned social expectations for masculinity or femininity, importantly enables individuals to participate in society and find acceptance from those witnessing and engaging in their gender portrayal. Over time, learning, internalizing, and displaying a socially accurate and acceptable mode of gender undergirds not just one's sense of self but also the way an individual is perceived and interacted with. Moreover, failing to provide such a gender portrayal bears social consequences, with individuals being rejected, harmed, and marginalized for not abiding by normative expectations. Sex and gender thus become 'social facts,' with tangible consequences on relationships, intimate bonds, and identities (West and Zimmerman 1987). And as NIPT routinely reveals sex chromosomes (and related aneuploidies), with associated assumptions about a future child's gender, societal tides around understanding reproduction through the fabric of gender are ever more prominent as early as 9 weeks into human reproduction.

In addition to making sex chromosomes more visible, prenatal genetic technologies like NIPT pronounce related genetic variations – classified under the diagnostic category of an SCA, wherein an individual 'strays' from the typical medical expectation of having XX or XY sex chromosomes. Identifying these differences, despite their relative lack of phenotypical presentations, is not inconsequential. Daniel Navon (2019) discusses this as the lasting repercussions of a 'genomic designation.' Attaching a genetic difference to a medicalized genetic condition produces not only new categories for diagnoses but rippling consequences for health and social identities. A genomic designation refines and reinforces the boundaries between normalcy and pathology: "knowing that someone has a mutation can recast otherwise normal observations as pathological findings that require further evaluation and perhaps treatment (Navon 2019: 250, 273)." When it comes to prenatal findings, knowledge of a genetic difference changes how we understand the fetus and eventual child (if the pregnancy was continued) as at-risk and possibly abnormal, as scientific experts and concerned parents seek to make sense of a variance so that it may fit their expectations for a diagnosis (Navon 2019: 271). Regarding SCAs, which often have little to no health implications or bearings on socially established sex and gender identities, this can mean processing the fetus or child through a pathological existence with heightened medicalized surveillance, when indeed the genetic difference may never have substantially shaped their lives at all. In other words, simply having a biomarker – an identified genetic difference – can lead experts and parents to "see a presentation in a person with a genetic mutation where they otherwise would not," and

pathologize the individual's existence per a deviation from normative expectations as they seek to explain the consequences of this difference (Navon 2019: 253).

Genomic designation is powerful in SCA prenatal findings, as elevating this diagnostic category has important social consequences for our collective understanding of sex and gender. NIPT has fundamentally altered how we reframe sex and gender in terms of medical pathology. Unlike prior prenatal genetic technologies and clinical practices, NIPT ('gender test') routinely screens for sex chromosomes as a part of the genetic markers it looks for, bringing information about a fetus' combination of Xs and Ys as well as their perceived eventual social identity into the broader medical understanding of the pregnancy's health and normalcy. Before NIPT expecting parents may have relied on ultrasound findings of genitalia and gonads to conceptualize fetal sex; now, NIPT illuminates sex chromosomes as a part of the constellation that comprises a medical classification of binary fetal sex, establishing more opportunities for parents and providers to 'see' discordance between a fetus' sexed features. And despite this discordance – e.g., a fetus presenting with a Y sex chromosome while resembling female genitalia – rarely having substantial health implications, knowledge of such genetic variation shapes how parents and providers understand the normalcy of a pregnancy, resulting reproductive decisions, medical surveillance and interventions, and social perceptions of what it means to embody a socially acceptable sexed and gendered existence.

Here, we see the consequences of genomic designation unfolding; while the SCA may not actually present a medical problem, in order to interpret the genetic difference, patients and providers collectively pathologize 'discordant' sexed features or atypical sex chromosomes in light of social assumptions around binary sex and its supposed interrelation with cisgendered identities. Providers, being reproductive physicians and genetic counselors in clinics as well as physician-researchers at conferences, co-productively pathologize such existences as deviations from normative binary sex and typical gendered expectations, often framing medical interventions as necessary to align such individuals with appropriate presentations of what it means to be a boy/man or girl/woman in society. This pathologizing is perhaps unintentional but occurs nonetheless as providers discuss SCAs in terms of normative sex and gender expectations with patients. This pathologizing also extends the medical gaze to various biophysical traits or characteristics that may have never otherwise been medicalized issues requiring intervention, certainly not at the prenatal stage. With the genomic designation of an SCA, infertility, height, and mild learning or behavioral challenges become reframed as medical concerns in-utero (rather than relatively straightforward issues that may be addressed if and when they emerge during one's life course), and moreover as relevant considerations for how a pregnancy is understood as normal or desirable.

Sex and gender not only become more relevant as medical categories at the earliest stages of human reproduction, but they are evermore intertwined with societal conceptions of normalcy and desirable existence. As normative expectations around sex and gender are elevated via genomic designation and pathology, these socially biased notions are imposed as medical facts onto a fetus that has not had a chance to exist among others let alone develop a sense of self. It is important to recognize the power that medical and scientific experts hold as they co-produce the pathology undergirding SCAs based on their understandings of normal sex and gender. Experts bring authority to claims around what makes for abnormal sex and gender existences, determining not just medically but also socially how men or women *should* appear and behave (Gonsalves 2020).

As medical experts compress and conflate their subjective social biases on sex and gender into making 'objective' facts, broader societal ideas around normal or acceptable existences are gradually constricted into reductive and exclusive sex and gender categories. And as the medical boundaries around who represents a normal versus perverse existence are increasingly sharpened in the process, there are lasting social consequences for how we collectively accept, include, and respect diverse ways of existence and expression.

This pathologizing and shrinking of how we understand appropriate modes of sex and gender is taking place at the same moment that scientific research shows that sex is not a binary category and need not correlate with gender identities. The idea of binary sex has long been shown as overly simplistic and reductive, with scientists illuminating how the gamut of biophysical features – genitalia, gonads, secondary sex characteristics, hormones, cellular structures, genes and chromosomes – can present with varying sexed identifiers, not all of which correspond to one another. Rather than expecting continuous alignment on these biomarkers, the science around sex shows that mosaicism or chimerism is not only common but perhaps to be expected in human variation (Ainsworth 2018). Still, that SCAs are being increasingly pathologized in terms of narrow sex and gender norms highlights the power of social assumptions in pervading and grounding what we collectively accept as scientific fact leading to medical diagnoses.

In the prenatal context, parents often grapple with their expectations for a to-be child, their family dynamics, and the child's future social relationships within the framework of sex and gender. They consider how they might raise a child who is different from those whose sexed features fall neatly into a male/female binary and what this might mean for how the child expresses their gender identity as potentially deviating from their embodied characteristics. As they navigate the uncertainty and presumed gendered implications around SCA diagnoses, parents' concerns are reflected in further medical interventions during pregnancy as well as decisions to terminate. While this data suggested that there are no obvious patterns around termination for SCAs, a notion also supported in broader population data, prenatal findings indicating SCAs are nevertheless increasingly impactful on termination decisions. One study, reflecting reproductive decisions following prenatal genetic testing during the 1970s and early 1980s, showed that out of 281 pregnancies diagnosed with an SCA via amniocentesis, 67.3% were terminated, and of 17 pregnancies diagnosed with an SCA via CVS, 88.2% were terminated (Navon 2019: 293). More recent data shows these figures around termination for SCAs remain relatively high, despite often no observed issues on ultrasound. A meta-analysis of studies between 1987 and 2004 revealed that 20-54.5% of expecting parents terminated for an XYY finding while 17-70% terminated for XXX, even though these pregnancies did not present with other anomalies or structural concerns (Hamamy and Dahoun 2004). Further, researchers at the University of California San Francisco found that 65% of expecting parents who learned their fetus had Turner Syndrome terminated their pregnancy, as did 70% of those whose fetus was diagnosed with XXY, 57% whose fetus had XXX sex chromosomes, and 40% who were found to have XYY (Shaffer et al. 2003). Despite SCAs having rather promising medical prognoses and extant supports (and at times no observable phenotypical differences at all), termination rates following such findings remain rather significant, highlighting the non-negligible consequences of expecting parents having to more frequently grapple with heightened uncertainty and complexity in their pregnancies.

With prenatal genetic screenings like NIPT identifying sex chromosomes, and thus SCAs, at greater frequencies, one can imagine the rates around termination for these conditions increasing. Acknowledging this possibility, the Association for X and Y Chromosome Variations (AXYS), the main advocacy and support organization for those with SCAs and their families, emphasizes the need for genomic education and support for expecting parents who learn of an SCA prenatally. AXYS aims to persuade expecting parents against terminating pregnancies due to SCA findings, reinforcing that these conditions are mild and variable, and that people with SCAs go onto lead successful social, professional, and personal lives. The group supports the need for early identification and treatment of SCAs, while underscoring that many individuals with SCAs are not notably impacted by their genetic variation and may not need substantial medical interventions ("AXYS" n.d.). As NIPT continues to be implemented ubiquitously, with plans for more integration into public prenatal care approaches, it remains to be seen how AXYS' objectives will fare against the increasing pathological framing of SCAs within medical contexts. The life experiences of those who live with SCAs, and social perceptions of these individuals as 'abnormal,' hang in the balance.

Reflecting on how genomic designation "may lead to lowered expectations, self-fulfilling prophecies, overtreatment, and perhaps even the reproduction of social inequalities," Navon (2019) writes that we "need to grapple with the way knowledge about mutations is actually being used in practice before we stumble into new versions of old tragedies […]. We need to think carefully about what it means to judge human difference based on what we know, or think we know, about a combination of their family backgrounds and the genetic mutations they bear (Navon 2019: 271)." The "old tragedies" he refers to are those of eugenics practices systematically seeking to eradicate and ostracize individuals and communities whose genetic and social backgrounds do not align with white supremacy and class-based elitism. Such concerns certainly apply to the case of NIPT elevating SCA diagnoses as genetically marked perverse sex and gender existences. Along these lines, we need to consider the consequences of a medical classification. Especially for SCAs – which bear little to no significance on how one would otherwise experience their health, personal, and social identity had they not known about their chromosomal variation – it is worth questioning whether applying a diagnostic label to such differences, designating them as medicalized conditions, is meaningful to affected individuals and their families. As this chapter as illustrated, it is often perhaps more beneficial to patient care, reproductive decision-making, and broader social equity when differences that have no clinical implications are not pathologized.

There are amplified consequences to establishing a diagnosis based on pathologizing fundamentally social conceptions, such as sex, gender, and sexuality, which influence how we perceive ourselves and each other. Not only must diagnosed individuals, regardless of realized impacts on their health, process their identity through a medical lens of abnormalcy, but their community may also recast this person as 'different,' or 'deviant' based on increasingly rigid boundaries around acceptable sex and gender identities. Even if these diagnostic foundations and assumptions are eventually debunked – as they have been with the 'criminal chromosome' – there are pernicious lasting effects to having established these ideas as scientific facts. Once brought to light, these conceptions have lingering and socially severe consequences, as they continue to circulate in the public imagination and often make their way into medicalized spaces, which in turn lends authority to the possibility that the myth may indeed have credence. As such, once again, diagnostic labels and related pathology do not always bring medical benefit and at times do greater

social harm. On a societal level, narrowing expectations around who we view as embodying a meaningful or appropriate existence, based on supposedly scientific notions of normalcy, shape both the types of individuals we choose to reproduce and the quality of life for every member of society. As variation among humans remains consistent, regressive social prejudices being reinforced as biological truths will undoubtedly constrain our views on disability, difference, and deserving existence. Difference goes from being a cherished value that enhances communities, to one that is unwanted, precluding the diversity that adds to our social and personal experiences.

While this chapter has critiqued NIPT in the context of SCAs, NIPT can be an incredibly useful technology for prenatal care and reproductive decision-making. Not only is it becoming increasingly accessible for patients, but it allows a low-risk, non-invasive approach to screening for possibly severe chromosomal conditions such as Down, Edward, and Patau Syndrome. This information helps parents to make reproductive decisions aligned with their values and enables more attentive and prepared clinical care among providers. For life-threatening conditions like Edward and Patau Syndrome, early prenatal screening can also spare parents more grief of a miscarriage or still birth, or of witnessing their infant suffer or pass soon after birth. In contexts like the United States, where disability supports are lacking and healthcare can be extremely costly, being able to exercise such reproductive choices is not only empowering but may also be pragmatic for families. Given these benefits of NIPT, it is important to consider how the technology can be harnessed more effectively, compassionately, and with more attention to optimizing its advantages.

Similar to other commercial genomic technologies in the United States, NIPT faces the pitfalls of a tool that was marketed too early and too quickly to consumers and clinics, without adequate attention to patient-centered clinical benefit and societal equity. Based on this data and the context of NIPT as used for conditions like SCAs, I provide three recommendations for how the screening tool can be implemented more critically.

First, prenatal genetic screening needs to focus on genetic differences and conditions that have evident health implications in-utero or necessitate medical interventions soon after birth. There needs to be a tangible clinical benefit to identifying and diagnosing a genetic difference prenatally, as opposed to medical interventions that are grounded in aligning individuals with socially biased views of normal existence. NIPT practices should focus on screening for differences that readily present phenotypical concerns in the fetus, which are clinically beneficial to ascertaining the pregnancy's health, and can empower a range of reproductive decisions and provider preparations.

Second, there ought to be more genomic education for parents prior to testing. Every expecting parent needs to have systematic access to thorough pre-test counseling with a licensed genomics provider where they can learn about their prenatal genetic testing options, how prenatal genetic tests function as medical tools (rather than as a "gender test"), and the types of results (and ambiguity) they can expect.

Third, more customized optionality around what expecting parents may want to use NIPT to screen for based on their values would be useful. After patient education, pregnant parents should be able to consider and select what information they would like to learn about via NIPT, having understood possible conditions that may be identified, the range of associated phenotypes, the probability-based nature of results, chances of false positives, possibility of mosaic or inconclusive results, the

chance that the finding may not be derived from the fetus at all, and that some findings may not have immediate clinical implications for their pregnancy. Allowing for more choice in NIPT screening panels moves the technology's implementation from being routine and uncritical to being a tool for personal reproductive empowerment aligned with individuals' varying views and values. Even with the option for customization, perhaps standard genetic screening panels should be limited to genetic conditions that have realized health implications that would change prenatal management, immediate birth needs, and post-natal care. These conditions might be those that are medically consequential, which would *require* a fetus or child to undergo interventions in order to enable survival, or those that lead to health challenges that substantially compromise one's life quality and longevity in the United States, wherein parents may not be able or willing to accept such challenges. In this way, NIPT can become framed more as a medical tool, providing information that is valuable to ascertaining health and reproductive choices, rather than a 'gender test' or a screening that returns results unnecessarily complicating parents' reproductive experiences and providers ability to provide high-quality prenatal care.

Currently, the use of NIPT toward identifying SCAs prenatally does not reflect that of a medical tool implemented fundamentally for health benefits. Medicalizing SCAs based on sex and gender pathology assumes that concordance between biophysical features like sex chromosomes, genitalia, gonads, and subsequent determinations of gender identities is needed for normalcy, healthiness, and social acceptance. Defining a diagnosis this way implies that anyone outside these limited bounds will be socially marginalized based on outdated notions of genetic essentialism, binary sex, and embodied gender. This is reflected in 'treatments' for SCAs, which are framed as medicalized interventions to align individuals with narrow ideas of 'normal' masculinity or femininity, rather than more nuanced sociomedical discussions about the need for greater tolerance and respect for genetic differences. As a result, medical authorities (even inadvertently) are supporting a return to socially regressive and unscientific ideas about genetic essentialism and biological determination, with ripple effects for patients' understandings and the public imagination around genetic variations and implications for 'normal' or desirable existences. With the sharpening of how normal versus pathological is defined, based on social prejudices being recast as clinically relevant diagnoses, there is a perpetual narrowing of our values around celebrating diverse existence. Given the power behind genetic tools like NIPT, and the credibility these tools gain when they are implemented by medical and scientific practitioners, it is of utmost importance that we approach their clinical use with caution, tempered and selective applicability, and thoughtfulness toward not only patient-centered experiences but the consequences for reinforcing and amplifying perilous opportunities for systematic social exclusion and inequity.

CHAPTER FOUR

Turf Wars in Reproductive Genetic Medicine: How Doctors and Genetic Counselors are Reshaping a (Feminized) Expert Role

The field of medicine is laden with contentions over professional jurisdiction. In the United States, mid-level or auxiliary healthcare providers, including nurses, physician assistants, technicians, and midwives have consistently sought more professional autonomy and recognition for their work in comparison to the more authoritative and well-recognized role of physicians. These professional groups have challenged, with differing degrees of success, their scope of practice, licensing requirements, supervisory dynamics, ability to order and interpret diagnostic tests, and use of technologies as they relate to physicians' roles (American Medical Association 2021; Black, Carlile, and Repenning 2004; Spilsbury and Meyer 2005). As these groups advocate to adjust the boundaries around their professional scope, their efforts remake the types of expertise and experts considered valuable within their medical fields. Enabling non-physician providers to expand their professional jurisdiction and be better recognized for their specific expertise (e.g., through inclusive billing codes, increased monetary benefits) is critical to uplifting the range of experts who contribute to patient-centered healthcare.

One such 'turf war' currently unfolding in the precision medicine field pertains to genetic counselors, who are vying for increased professional autonomy via the 2021 Access to Genetic Counselor Services Act (also referred to as 'HR2144,' the 'Act' or the 'NSGC Act'). Through this Act, genetic counselors – a professional group consisting largely of women – are seeking their specialized genomics expertise to be more valued and accessible, citing a rapidly evolving environment where patients need more dedicated support around genetic testing (Higgins 2021). The National Society of Genetic Counselors (NSGC) are advocating to expand counselors' ability to order and bill for genetic tests, which is currently limited to licensed physicians. They assert that enabling genetic counselors to order and bill for testing is not only warranted given that counselors are the experts interpreting these test results for patients, but also that doing so would critically augment patients' access to healthcare that increasingly relies on personalized insights from genetic tests to diagnose and treat medical needs (Higgins 2021). This is pertinent in prenatal care, where genetic testing via Noninvasive Prenatal Testing (NIPT) is commonly used to understand risks for various chromosomal trisomy conditions and identify fetal sex chromosomes. As NIPT and other reproductive genetic tools become more routine, and at times necessitate further prenatal genetic tests, there is a growing demand for prenatal genetic counselors' expertise. Moreover, there is an opportunity for genetic counselors, a historically feminized profession, to reshape the types of knowledge and expertise considered valuable in this space.

New technologies like NIPT also shape expertise, expert roles, and the relationship between physician and counselor groups (Barley 1986). As new genomic technologies are incorporated into routine clinical practices, experts contend over how these tools should be used, by whom, and toward which healthcare goals. These debates are shaped by concurrent sociohistorical moments that inform changing priorities in healthcare. Most relevant here is the shift toward precision medicine, which relies increasingly on individualized genetic insights to manage one's clinical care, as well as the ongoing emphasis on personalized patient-centered healthcare models, which underscores the need to recognize each patient's psychosocial context (i.e., feelings, concerns,

values) as well as to develop meaningful provider-patient partnerships so that medical processes are concordant with patients' values (Epstein et al. 2005; Mukherjee 2020). In reproductive medicine, providers and patients increasingly rely on prenatal genetic testing, such as NIPT or amniocentesis, to help make reproductive decisions and medically manage pregnancies (Clarke et al. 2003; Minear et al. 2015; Timmermans and Kaufman 2020).

However, because NIPT (like other genetic technologies in the United States, e.g., genetic ancestry and health testing, polygenic embryo screening) was initially introduced through the consumer health market, rather than via clinicians, the medical 'best practices' around how the tool should be used have lagged behind its on-the-ground implementation (American College of Obstetricians and Gynecologists 2021). As a result, there are inconsistencies across clinicians' practices with regard to the recommendations for how prenatal genetic testing using NIPT should be approached compared to how it is practically carried out (van der Meij et al. 2022). At a few practices, patients are able to see a genetics provider before they pursue prenatal testing to discuss each testing option in-depth. Many other practices with limited genetic counseling resources rely on non-genetics physicians (i.e., Obstetrician Gynecologists) to explain NIPT to patients, which they often do incompletely or incorrectly. Consequently, patients at some clinics may never see a genetic counselor for a thorough explanation of testing (unless their NIPT results suggest possible concerns), and some may be unaware they have provided a sample for genetic testing because their blood was drawn as a part of other standard pregnancy screenings. In this way, patient management between physicians and genetic counselors can be challenging. As physicians hand-off patients with concerning or complex test results to genetic counselors, often without a full and accurate understanding of prenatal genetic testing, counselors are left to 'back track' and mitigate patient misconceptions ('damage control,' as they describe it).

While new technologies can raise issues around providers' workflow and patient care, these challenges also create new opportunities (Barley 1986). As expert groups more frequently engage with technologies like NIPT, given its routinization in prenatal care, they collectively define best practices, change perceptions around each other's expertise, and reconfigure professional jurisdictions to meet emerging patient needs. As a result, in the case of prenatal genetic medicine, we see how genetic counselors transition from being auxiliary providers to central genomics experts needed for equitable patient care. And as genetics becomes more integral to medical care in the precision medicine era, we can expect the genetic counselor's role to become more salient. The Act and the on-the-ground shifts in expertise between doctors and counselors discussed in this chapter mark the budding possibility of a larger transformation in this field, with counselors, their expertise, and their specialized approach to genetic medicine becoming more significant.

Background on the "Access to Genetic Counselor Services Act"

The "Access to Genetic Counselor Services Act" (HR2144) provides an important backdrop to understanding the relationship between reproductive physicians and prenatal genetic counselors – the two key groups of experts engaged in prenatal testing and care. It highlights the changing dynamics around the expertise needed for patients in this moment of routinized prenatal genetic testing. The NSGC helped put forward the Act to the US House of Representatives in 2021. Framed as an effort to "modernize" Medicare for the genomic (or precision) medicine era, HR2144 aims to recognize certified counselors as providers under the Centers for Medicare and Medicaid Services (CMS). Specifically, the Act would "provide reimbursement to genetic counselors at 85

percent of the physician fee schedule amount" and improve other practitioners' (e.g., physicians) ability to refer patients to counselors. If passed, counselors would be able to directly order genetic tests and bill for their counseling services (Higgins 2021; National Society of Genetic Counselors 2022).

The NSGC, their sponsors, and signatories supporting HR2144 emphasize that these measures are a matter of equity in public healthcare access, given that genomic medicine is becoming increasingly prevalent.[50] Stakeholders problematize the current "arcane" Medicare coverage, where genetic counseling is a covered benefit (and often required to order certain genetic tests), but counselors themselves "cannot be reimbursed for providing services to Medicare beneficiaries, which impedes patients' access to these uniquely trained healthcare professionals" (National Society of Genetic Counselors 2022). They note that "lack of access (to genetic counselors) can result in harm such as incorrect interpretations of genetic test results, failure to identify individuals who have increased genetic risk, and inaccurate risk assessments leading to inappropriate medical management and sometimes death" (National Society of Genetic Counselors 2021b). Thus, direct access to genetic counselors is framed not only as an improvement in public healthcare quality and accessibility, allowing "patients (to) receive the counseling they need to make the right decisions about their care," but also a measure to "save money for beneficiaries and the Medicare program" by ensuring other providers appropriately order and interpret genetic tests (National Society of Genetic Counselors 2022).

As they attempt to position themselves as "qualified Medicare providers," genetic counselors emphasize that their specialized expertise and knowledge is best suited for the growing demands of precision medicine. In support letters, media releases, and the Act itself, genetic counselors underscore they are "highly trained" and "specialized" providers with "advanced training in medical genetics and counseling," which enables high quality patient education on "both medical and psychological" fronts. In highlighting the counselor's role for patients and their families, as well as their integral function within medical teams, the Act uplifts the need for holistic, socioemotionally attentive, and culturally competent patient-centered care when handling genetic tests. It stages a moment where genetic counselors are mobilizing their specific expertise to make the case for a new type of expert provider needed to adequately serve patients in a quickly developing era of precision healthcare. Mobilizing to make genetic medicine more equitable and attentive toward patients' needs is particularly critical given the history of eugenics that grounds precision medicine; while the Act may not be an all-encompassing solution to genetic medicine's fraught roots, these efforts ensure that the field contends with its history and expert capacities (Phillips 2020). (Higgins 2021; National Society of Genetic Counselors 2021b, 2022).

For their part, physicians have taken varying stances on the HR2144. Almost all the individual physicians I interviewed expressed some degree of support for the Act. However, as a whole, the American College of Medical Genetics and Genomics (ACMG) – the main professional association of doctors in the genomics field –initially opposed counselors' efforts, drawing sharp

[50] This Act has been sponsored by U.S. Senators across partisan lines each time it has been introduced into Congress. Those who have sponsored the Act include Senators Jon Tester (D-Montana) and John Barrasso (R-Wyoming), and Representative Brian Higgins (D-New York). The Act was created in close collaboration with NSGC, the central voice in mobilizing for these efforts. Stakeholders including genetic counselors, patients and their families, genetic counseling and medical students, and physicians have provided letters to Congress members in support of the Act.

criticism from medical and patient communities for their gatekeeping attempts (American College of Medical Geneticists 2021). While ACMG's membership includes physician geneticists and other professionals working in medical genomics (in addition to some genetic counselors), their leaders largely represent physicians' interests. In January 2020, ACMG's then president and its CEO stated, "the ACMG cannot support any policies that would permit genetic counselors to practice medicine," which they considered to include ordering genetic tests. They framed the Act as contrary to best practice patient care, in that it would allow non-physicians 'too much' independence; the ACMG's stance on patient care necessitated physicians every step of the way. As they defined their "practice of medicine," insisting that non-physicians like counselors were not suited to delivering medicine this way, the ACMG sought to limit counselors' scope to require supervision by physician geneticists and prohibit counselors from "independent ordering of genetic tests […] as well as interpretation and return of results to patients without any interaction with physicians" (American College of Medical Geneticists 2021; Raths 2020).

Pushing back on the ACMG, stakeholders supporting NSGC noted that direct supervision of every genetic counselor is "neither necessary nor feasible" given that the medical geneticist (physicians specialized in genomics) workforce is much smaller and growing less slowly than that of genetic counselors. Moreover, they underscore that counselors, with their specialized skills, currently advise non-geneticist physicians on which genetic tests to order and how to correctly interpret results, precluding the need for constant direct supervision. Since receiving wide disapproval for its stance, the ACMG has clarified its statement (Raths 2020).[51] More recently, they proposed that the Act's language be modified to require "collaborative agreements (NOT supervision) between counselors and physicians," that can be tailored and customized per their institutional needs (American College of Medical Geneticists 2021). ACMG's stifling of NSGC's aims has engendered tension between the two professional societies over not only expertise but what it means to be a medical provider in the genomics era.

HR2144 is gaining support among medical societies, lawmakers, and healthcare organizations, possibly positioning more experts (i.e., genetic counselors) as central to the genomic medicine forefront (National Society of Genetic Counselors 2022; Ray 2020). In reproductive medicine, where genetic testing is increasingly commonplace, the Act presents an important sociohistorical moment bringing to the surface claims around which types of expertise are valuable and best suited to meeting patient needs within precision medicine. As counselors negotiate their roles, potentially transforming their professional jurisdiction and relationship to physicians, they challenge the established order of expertise in genomic medicine. Especially given that the overwhelming majority of genetic counselors are women, representing a historically feminized profession, their efforts not only confront medicine's traditional gatekeepers but also stand to reconfigure the types of knowledge and expertise valued in prenatal medicine to reflect skills typically considered 'feminine.' Through this Act, genetic counselors could reshape the genomic medicine field, uplifting feminized expertise that has been traditionally less recognized and de-valued.

The Need for Genetic Counseling Experts

[51] Genetic counselors criticized ACMG for undermining their critical contributions to a medical team and patient care, and misleadingly framing their HR2144 efforts as aiming to 'practice medicine' (e.g., the Act does not state that genetic counselors should be able to provide diagnoses and treatment, which they argue constitute 'practicing' medicine) (Raths 2020)

The demand for genetic counselors emerged in the 1960s, following a decade of advancements in medical genomics and genetic technologies. Between 1956 and 1966, researchers found that the typical human DNA consists of 46 chromosomes, showing that Down's syndrome resulted from a third 21st chromosome, and identified numerous other chromosomal conditions, including sex chromosome aneuploidies like Turner and Klinefelter syndromes. The 1970s also saw the emergence of prenatal genetic diagnoses, with developments in amniocentesis and ultrasound. These transformations in healthcare were also taking place against the background of feminist health movements to make abortion and contraception more accessible (Minna Stern 2012). These developments called for a group of experts (i.e., genetic counselors) who could "reliably translate (genetic) test results and technological language for a diverse clientele," including patients, physicians, and scientific researchers, aiming them with equal doses of scientific acumen and human empathy to make decisions about their options" (Minna Stern 2012:1). Particular to prenatal screening, "counselors' overriding objective is to help parents make autonomous decisions about screening, diagnostic testing, possible preparation for the birth of a child with disabilities, or pregnancy termination" (Minna Stern 2012:15). Given that sessions often vary based on the type of test, diagnosis, and sociocultural backgrounds, among myriad other factors, professional societies like the NSGC suggest that "genetic counseling sessions should consist of informed, educational consultations that are fluid and responsive to the genetic condition and personal circumstances at hand" (Minna Stern 2012: 9, 14).

Reflecting these expertise needs, genetic counseling formally culminated into a profession in 1969, with the first genetic counseling Master's program established for women at Sarah Lawrence College. Earning a Master's degree in genetic counseling, where students specialize in genomic science and targeted counseling strategies across medical specialties, in addition to completing various licensing and certification qualifications, remains a requirement for genetic counselors today. Following Sarah Lawrence, several subsequent programs were established in the 1970s, including at Rutgers University, University of California Berkeley, and the University of Colorado. These genetic counseling programs were highly gendered, aimed specifically at middle-class, white mothers, as the profession was deemed a suitable option for women to participate in the workforce. Genetic counseling was seen as a respected, yet auxiliary, healthcare position, apt for women seeking to work outside the home. It also involved relational and empathetic counseling skills, typically considered feminine work. Most importantly, counseling could be performed on a flexible, part-time basis, leaving room for mothers to fulfill their primary household and childcare duties at home. These decade-old gendered patterns persist in the largely feminized field, which continues to lag in terms of its professional diversity; as of 2019, 95% of genetic counselors identified as women and 90% as white or Caucasian (National Society of Genetic Counselors 2019). Today, the feminization of this profession contextualizes how genetic counselors are seeking better recognition and compensation for their skills and expertise; their efforts could reallocate value to expertise typically considered feminine and underappreciated. (Minna Stern 2012)

With the Human Genome Project (1990-2003) continually facilitating innovations in medical genetics (i.e., cancer screening, gene therapies, non-invasive prenatal screening), genetic counselors are more pertinent to healthcare than ever. Although the profession is expected to grow more rapidly than other occupations – with 26% growth projected for genetic counseling compared to 14% for other healthcare roles and 8% for all occupations – there are currently only between

2500-4000 licensed counselors in the United States (Bureau of Labor Statistics 2022; National Society of Genetic Counselors 2021a). The profession is notably underrepresented considering the sharp increase in genetic tests conducted; where Medicare covered around 627,000 genetic tests in 2016, just three years later in 2019 they covered almost 2.1 million tests (235% increase) – not including tests paid for by private insurances or personal finances (Office of Inspector General 2021). Ideally, each person considering a genetic test would be able to weigh their decisions and results with a genetic counselor; however, many patients in the United States do not receive such counseling, and there are numerous challenges regarding how counselors can bill for their services and be accessible to patients via insurance coverage. The need for access to counselors is more visible as the NSGC pushes its Act forward, illuminating tensions among experts, their expertise claims, and the consequences for patient care in precision medicine.

These implications could be especially felt in the rapidly growing reproductive genetic testing space. Non-invasive prenatal testing (NIPT or cell-free DNA) accelerated prenatal testing, enabling detection of fetal genetic conditions as early as 9 weeks in a pregnancy using a standard blood draw from the pregnant person. Although these findings must be confirmed via diagnostic testing (e.g. CVS or amniocentesis), NIPT detects several chromosomal trisomy conditions (e.g., Down, Patau, Edward Syndromes) with high sensitivity and specificity (e.g., detects Down Syndrome with 99.9% sensitivity and 98% specificity) (Norton et al. 2015:1598). The reliance on genetic tests during reproduction is increasingly supported by medical organizations, including the American College of Obstetricians and Gynecologists (ACOG), who in 2020 recommended NIPT be offered to *all* pregnant people regardless of age (American College of Obstetricians and Gynecologists 2021). In the United States, 50% of genetic counselors practice in reproductive medicine – 30% in prenatal and 20% in preconception (National Society of Genetic Counselors 2021a). Thus, reproductive testing illuminates expertise claims and ongoing tensions that could be informative for other medical fields similarly routinizing genetic testing (e.g., cancer, pediatrics).[52]

The Emergence of Less Powerful Groups and Their Expertise

To understand how genetic counselors and doctors are transforming the boundaries around their roles and expertise, I first unpack what defines a profession, an expert, and related expertise, as well as how less powerful expert groups can emerge. Genetic counselors' efforts to broaden their scope of practice and emphasize their contributions to healthcare in many ways represents Andrew Abbott's (1983) conceptualization of professions per their necessary "service to society" (p. 855). Abbott describes that "professions take their shape from a series of *socially defined problems* of order *which their knowledge permits them to control* (Abbott 1983:878; *emphasis added*)." In order to solidify their professional jurisdiction, practitioners undertake efforts to define their specific

[52] Genetic testing during reproduction is particularly visible in California, from where this data is drawn, which has long been a "hub" for cutting edge reproductive services (Dunn 2019). The state was an early adopter of prenatal genetic testing, with the 1986 California Screening Program that pledged state-wide access to maternal serum alpha-fetoprotein (MSAFP) testing. Today, the California Prenatal Screening Program covers almost all major prenatal genetic tests, including the more recent NIPT, allowing wide implementation of these technologies across medical institutions and diverse patient groups (California Department of Public Health 2023). As such, California's reproductive medicine experts consistently engage with how these innovations impact their relative expertise, workflow alongside other experts, and approaches to patient care. Physicians and genetic counselors in California contend with considerations at the forefront of genetic testing, which can be more widely applicable as prenatal genetic testing gains similar ubiquity nationwide.

knowledge base and the significance of their social contributions (Abbott 1983: 856, 865). For genetic counselors, we see this historically, as graduate programs to train professional counselors evolved alongside developments in genomic technologies around the 1960s to meet the needs of providers and patients newly using these innovations. Further, because professions arise to meet socially defined problems, groups often need to redefine or augment their professional boundaries based on shifting societal contexts that change the significance of their roles and contributions (Abbott 1983: 877) – much like genetic counselors are doing through the NSGC Act, given how their profession is specifically attuned to meeting patient needs in the current precision medicine era where genetic testing is increasingly routinized in clinical care. Reliance on genetic testing insights to enable medical diagnoses, treatments, and preventative measures has not only heightened the demand for professionals with specialized knowledge in genomic science but also calls for those with the ability to appropriately communicate these complex results to patients and other providers. These transformations in our approach to medicine, Abbott (1983) might say, point to a moment where genetic counselors can rewrite the boundaries of their profession to better capture their increasing pertinence to patient-centered care in genetics.

While useful in defining professions and professional groups, Abbott's (1983) theory emphasizes how established or powerful groups, such as doctors, can determine professional jurisdictions, leaving the question of how less powerful groups (e.g., genetic counselors) emerge under-explored. For the latter, we can turn to Gil Eyal's (2013) work on experts and expertise. In particular, Eyal (2013) highlights how non-experts—with less power and less established jurisdictions compared to experts (or professionals)— can gain significant influence over a field via claims to specific expertise (Eyal 2013: 899). Here, expertise is a larger apparatus, "linking together agents, devices, concepts, and institutional and spatial arrangements (Eyal 2013: 863)." In Eyal's case, the widespread historical deinstitutionalization of those with mental illnesses enabled a sociomedical context where non-experts (e.g., "parents of children with autism in alliance with psychologists and therapists") could make significant claims to expertise in ways that fundamentally altered medical trajectories for children with autism (p. 863).

Substantial other research has bolstered this idea that non-experts, or less powerful groups, can have significant influence over a medical issue through various claims to expertise that resonate with their respective contemporary sociomedical contexts. Stephen Epstein illustrates the centrality of patients and their allies in urging a response to HIV/AIDS (Epstein 1995), while Alondra Nelson highlights Black Panther activists' role in national sickle cell interventions (Nelson 2011). Specific to reproductive medicine, Rima Apple shows how mothers and health advocates as early as the 1930s and 1940s mobilized against medicalized recommendations misaligned with their pregnancy and post-natal experiences (Apple 1995, 2014). In genomics, Navon and Eyal illuminate patient advocacy as key to developing diagnostic classifications for conditions including autism, DiGeorge, and Jacob syndromes (Navon 2019; Navon and Eyal 2016); others have illustrated how patients and their families provide key expertise that makes genomic data clinically meaningful (Markens 2013; Timmermans and Buchbinder 2012; Timmermans and Kaufman 2020; Timmermans and Stivers 2017). In each instance, less powerful, non-expert groups have had notable impacts on medical fields through their unique claims to expertise and have been able to define their significance as a group accordingly.

Where most of the literature on jurisdictional struggles centers gatekeeping efforts, scholarship on non-experts helps explain how genetic counselors as a less powerful group are gaining influence through boundary-remaking with doctors. While genetic counselors are not non-experts – they are most certainly an established group of experts with a professional jurisdiction – in relation to reproductive physicians, they have considerably less professional power, authority, and recognition of their expertise. Indeed, these are some of the fundamental issues that the NSGC Act is aiming to address. This chapter shows how genetic counselors are augmenting their jurisdiction and influencing the genomics medicine field via their claims to expertise. As sociomedical contexts (e.g., deinstitutionalization in Eyal's case) are crucial to enabling less powerful groups' expertise claims and influence on a field, genetic counselors are similarly propelled by the current moment. The ongoing routinization of genomic medicine (our shift toward precision medicine), as well as the sustained movement around patient-centered care are critical to shaping how genetic counselors make specific claims to expertise and position themselves as integral to current patient needs. In the current sociomedical context, genetic counselors could cement themselves as a central healthcare provider, transforming the experts and expertise defining genomic medicine.

How Doctors and Genetic Counselors Shuffle and Absorb Expertise

In order to explain *how* less powerful groups make claims to expertise and gain footholds within a field I turn to theories of action. Social action theories suggest that groups mobilize resources to achieve particular outcomes for themselves. Groups that are dominant within a field (e.g., physicians) often seek to maintain the status quo given that extant arrangements are beneficial to them; this was evidenced in the ACMG's initial resistance to the NSGC Act. On the other hand, challenger groups can disrupt or transform the 'rules' or structures of a field to enhance their roles or influence (Fligstein 2001:109; Fligstein and McAdam 2011). In addition to the NSGC Act, genetic counselors contribute innovative approaches to interpreting complex genetic information for clinical care, exemplifying their role as 'challengers' in this context (Markens 2013; Pollack 2012). These theories also underscore that 'moments of crisis' – like unmet demands in patients' equitable access to genomic healthcare – can create opportunities for a field to transform (Fligstein 2001:117, 123; Swidler 1986). Here, the reorganization of expertise among genetic counselors and physicians, alongside the backdrop of the NSGC Act seeking to augment counselors' professional authority, exemplify a moment where established schemas of practice in the genomic medicine field could be upended and revised.

The degree of success a dominant or challenger group achieves is dependent on their "social skill," which relies on the group's ability to strategically harness resources, mediate between groups, and create frameworks of cooperation to realize their claims (Fligstein 2001:105–7, 112, 115; Sewell 1992). As genetic counselors mobilize, we can conceptualize their resources as "cultural health capital" – a "repertoire of cultural skills, verbal and nonverbal competencies, attitudes and behaviors, and interactional styles […] (that) may result in more optimal health care relationships (Shim 2010:1, 11)." Through their workflow alongside physicians, as well as the claims put forward in the NSGC Act, genetic counselors are demonstrating that their approach to navigating genetic tests and related patient counseling exemplifies the type of health capital needed for equitable patient care in precision medicine. Counselors are also garnering stakeholder support for their efforts to uplift their expertise and expert roles. In terms of the Act, we see this represented in efforts to align healthcare professionals, patient groups, and lawmakers on expanding counselors' scope of practice and professional recognition. In clinics, as we shall see in this chapter,

counselors and physicians seem to be cooperating, perhaps inadvertently, to reorganize their expertise around patient care in ways that define both groups' professional jurisdictions and highlight counselors' specifically relevant expertise.

However, rather than a typical (perhaps more adversarial) dominant versus challenger group dynamic, counselors and physicians in clinics are engaged in a dialectical process to reconfigure their workflow and expertise to meet patients' genomic healthcare needs. This process in part represents Josh Seim's (Seim 2017, 2020) concept of "burden shuffling." As various expert groups work toward a shared objective, Seim (2020) shows how they often transfer responsibilities to one another. As they burden shuffle, experts in one profession push tasks they consider undesirable onto those in others, claiming that their professional roles do not consist of doing this type of work (Seim 2020:21, 115–16). In this case, doctors burden shuffle responsibilities related to specialized genetic knowledge – such as more involved socioemotional counseling and particularly ambiguous or complex prenatal patient cases– over to genetic counselors. They 'de-medicalize' these responsibilities as specialty non-medical knowledge, emotional care, or administrative logistics that are peripheral to their 'practice of medicine.' Shuffling tasks considered too detailed, time consuming, or generally unrelated to their professional roles, physicians instead insist their focus needs to remain on 'objective,' clinical management of a prenatal patient's medical needs. In doing so, reproductive physicians are simultaneously defining their professional jurisdictions while shuffling tasks they deem outside these medicalized boundaries to genetic counselors.

For their part, genetic counselors strategically absorb these shuffled responsibilities, mobilizing what Shim (2010) may describe as their 'cultural health capital' (i.e. enacting their "social skill") to frame these squarely within their expertise. Genetic counselors 're-medicalize' these absorbed responsibilities as necessary to delivering equitable medical care and responding to patients' healthcare needs as genetic testing becomes routinized.[53] Counselors underscore that the current sociomedical context requires professionals like them, who have in-depth genomics knowledge, are trained in socioemotional counseling techniques, and are dexterous when it comes to navigating ambiguous genetic data or complicated patient cases. Thus, per counselors, supporting the delivery of genetic medicine, in part, means providing in-depth specialized genomic care, including the socioemotional care and navigation of ambiguity. As shuffling and absorbing enables counselors to uplift their value, they expand what expertise and experts constitute the apparatus around equitable medical care in reproductive genetics.

While the shuffling/absorbing process among doctors and counselors seems cooperative, they are not necessarily consciously collaborating. Rather, each group acts in their own professional interest, with the shared goal of supporting patients through prenatal genetic testing. Reproductive physicians are aiming to protect the boundaries around their expert roles, defining the objective, clinical management that they focus on, while genetic counselors seek to enhance and uplift the specific expertise that makes them valuable healthcare providers deserving of greater recognition (i.e., via insurance and billing policies). As such, it appears convenient (rather than intentional)

[53] Genetic counselors are not claiming to practice medicine. Rather, in their 're-medicalizing' of expertise during the shuffling/absorbing process, genetic counselors are framing their strategically absorbed responsibilities as crucial to (rather than constitutive of) providing genetic medicine to patients. They position their expert roles, and their approach to patient care, as critically supportive of the larger expert care apparatus in genetic medicine, rather than representative of practicing genetic medicine itself.

that the tasks physicians consider burdensome and outside their jurisdiction spillover to genetic counselors in ways that are strategic and beneficial to counselors' expertise and jurisdictional claims. While doctors and counselors may not be intentionally cooperating as they shuffle and absorb responsibilities, this dialectical process is still one that requires mutually supportive action from both groups. As a result, we not only see a reorganization of professional responsibilities between reproductive physicians and genetic counselors, but also a potential transformation of the counselor from an auxiliary provider to a more central expert in prenatal genetic medicine.

Gendered Professions and Knowledge-Making

The shifting dynamics between counselors and physicians – and the ways their respective expertise are valued as a result – is consequential especially because of the gendered systems contouring the reproductive genetic medicine field. Where genetic counseling arose as (and remains) a feminized profession, the physician profession has historically been dominated by class-privileged men with exclusionary practices aimed at maintaining this influence (Minna Stern 2012; Nakano Glenn 1992; National Society of Genetic Counselors 2019; Starr 1982). These gendered systems structuring professions are reflected in how (and by whom) knowledge is produced within that field, and which forms of expertise are valued accordingly. In fields such as medicine, operating along largely male-dominated practices, professional groups of women are typically responsible for 'care work' involving emotional management, compassion, and intimacy (e.g., counselors' socioemotional counseling) that is often under-recognized as expertise (Acker 2004; Hochschild 2011; Hochschild and Ehrenreich 2004; Pande 2010). Traditionally, this feminized work has been characterized as "low-paid, dead-end, unskilled, boring and highly-detailed" (Salzinger 2004:19), diminishing a profession's perceived prestige and influence within a field (Williams 1992). However, as dynamics between a field's experts transform, so can the gendered ideologies that undergird what counts as valuable expertise, possibly bringing forward new expert perspectives. In prenatal genetic testing, this can reshape how equitable medical care is conceived and practiced.

In scientific fields (e.g. medicine), feminist scholars critique how masculinized assumptions have defined 'objective' knowledge-making. They underscore that masculinized assumptions about science and objectivity are "unlocatable" (i.e., without sociohistorical context) and thereby not only "irresponsible" in their claims (Haraway 1988:583) but also precluding more complex, thought-provoking knowledge from standpoints outside the white, male status quo framework (Harding 1987; Hill Collins 1986). Importantly, these gendered systems shape *who* can embody expertise and produce knowledge. This is consequential for determining which experts hold influence over a field, its constituent objective 'facts,' and the underlying assumptions informing its practices. For example, in reproductive medicine, physicians and scientists historically positioned themselves as experts by framing women's generational knowledge about childbirth and childcare as 'antiquated,' while emphasizing their medical knowledge as scientifically objective and superior (Apple 1995, 2014). More recently, research shows that scientific literature about gametes, sex chromosomes, and reproductive processes is imbued with deeply gendered ideologies, reflecting the masculinized expert groups producing this information (Martin 1991; Richardson 2012). As such, rather than being unquestionable objective knowledge, biomedicine can reflect biased subjectivities and masculinized modes of making experts and expertise.

However, knowledge-making systems can transform, and the NSGC Act represents counselors' efforts to do so within the genomics field.[54,55] As contexts change, dynamics between expert groups can shift, and previously under-recognized groups can become more pronounced and influential (Fligstein 2001). In reproductive genomic medicine, the sociomedical context around relational patient-centered care and routinized genetic testing provides the opportunity for experts to disrupt existing gendered power structures.[56] As doctors and counselors shuffle/absorb expertise, this context can enable genetic counselors to situate their typically 'feminized' expertise as more valuable and influential to the reproductive genomics field than it was previously considered (tangibly, this could unfold via the NSGC Act, as counselors mobilize to revise insurance billing policies that currently disregard their expert roles). In particular, genetic counselors' socioemotional counseling and skills around navigating ambiguous genetic data are both increasingly necessary and valuable to address patients' needs as prenatal genetic testing becomes commonplace. As a result, traditionally under-recognized feminized experts and expertise could be gaining a foothold in shaping 'best practices' in prenatal genetic testing and equitable patient care. With genetic counselors (and their feminized modes of knowledge and expertise) becoming more centralized, we could see perspectives that have been historically excluded informing today's medicalized practices around equitable patientcare.

Transforming Experts and Expertise: The Case of Doctors and Genetic Counselors in Reproductive and Genetic Medicine

The following discussion illustrates how experts – doctors and genetic counselors – in reproductive and genetic medicine negotiate and reorganize their expertise and professional roles surrounding prenatal genetic testing and patient care. It shows how doctors and counselors, as distinct groups of experts with differing levels of jurisdictional authority, can work collaboratively rather than contentiously (albeit, unintentionally) to transfer and absorb responsibilities in order to meet the emerging demands of patient care in reproductive genomic medicine. Patient care demands reflect a contemporary sociomedical context wherein 1) genetic technologies (e.g., prenatal genetic testing via NIPT) are being increasingly routinized in reproductive medicine while 2) patient-centered care and advocacy – the result of a sustained medical movement to establish greater partnership between doctors and patients – remains paramount (Epstein 1995; Mukherjee 2020;

[54] Indeed, Harding, Hill Collins, and Haraway each posit modes of resistance and knowledge reclamation. They reinterpret 'objectivity' as a science that is 'passionately detached' yet situated in researchers' particular standpoints – locatable and tied to other "webbed connections" that can reveal multiple vantage points. Fundamentally, these scholars conceptualize the scientific method as being able to produce critical knowledge that is historically and socially contextualized (Haraway 1988; Harding 1987; Hill Collins 1986).

[55] There are several examples of gendered knowledge reclamation in practice. For example, as early as the 1930s mothers organized around key scientific publications (e.g., *Natural Childbirth* by obstetrician Grantly Dick-Read) to question doctors' medicalized views on reproduction that contested women's lived experiences. In the 1960s, the 'International La Leche League,' run primarily by women, also successfully countered doctors' bottle-feeding directives by uplifting rationales grounded in mothers' standpoints and experiences. This movement coined the phrase "breast is best," pushing back on the contemporary 'best practice' from doctors calling for parents to rely on formula feeding their infants. (Apple 2014)

[56] Current research suggests that genetic counselors are becoming critical to interpreting genetic data and biomedical information in ways that physicians did not previously consider (Minear et al. 2015; Pollack 2012), situating themselves as a "key new profession […] between the ascendant complex genetic knowledge and the lay public, patients, and other medical professionals" (Markens 2013: 303, 306).

Navon 2019). As doctors and genetic counselors dialectically define their roles and give meaning to their distinct expertise as it pertains to patient needs during prenatal genetic testing, we see how this sociomedical context helps uplift the role of the genetic counselor and the value of their expertise; this process not only enables expert groups to transform but brings previously underrecognized experts and expertise to the forefront of genomic medicine. The ongoing shift in how genetic counselors' expert roles and expertise are becoming centralized is particularly significant because it marks a moment where less powerful groups may be redefining what it means to provide comprehensive and adequate medical care in precision medicine. It characterizes an opportunity for a traditionally feminized professional group (i.e., genetic counselors), with skills historically considered 'feminine' and thereby undervalued in the medical institution, to have notable influence over the prominent and growing field of reproductive genomic medicine and the parameters around patient healthcare in this space.

The data presented below are drawn from 40 in-depth interviews, 20 with genetic counselors and 20 with physicians, all of whom were working in reproductive medicine in California. All the physicians were obstetricians and gynecologists, with 15 further specialized in maternal and fetal medicine (MFM). Interviewees also occupied varying career stages, ranging from early practitioners (1-2 years) to those with decades of experience or recently retired. They worked at a wide range of medical settings, including academic medical centers, genetics laboratories and biotechnology companies, community and non-profit hospitals, private clinics, and large general hospitals. These institutions were located across urban and rural areas, low-income communities, and affluent neighborhoods, ensuring a range of perspectives and experiences. The interview data is also supported by observations conducted at classes and trainings held at various genetic counseling Master's programs, as well as observations of patient-genetic counselor consultations at a large non-profit hospital in rural California.

The first part of the empirical discussion depicts the process of burden shuffling (Seim 2017, 2020) and absorbing between doctors and genetic counselors, respectively. It shows how doctors de-medicalize tasks that require 'specialized genomics expertise' as outside their purview and impractical for them to carry out given the constraints on their time – they have too many pregnant patients to see with too much other medical information to cover, making delving into the details of genetic testing and its implications unfeasible. In doing so, reproductive doctors 'shuffle' these tasks they consider undesirable and non-medical onto genetic counselors, claiming that responsibilities requiring specialized genomics expertise are too detailed or unrelated to their physician role of objective, clinical management – the pragmatic 'hands-on' medical needs and clinical interventions around a pregnancy (Seim 2020: 21, 115-116). In turn, genetic counselors 'strategically absorb' shuffled specialized genomics expertise into their roles, framing these as squarely within their expertise and professional training. They re-medicalize these responsibilities as core to supporting medical care in genetics, positioning their expert ability in specialized genomics as central to addressing contemporary prenatal patient needs. Indeed, responding to patients' questions and concerns around in-depth prenatal genomics issues (e.g., types of tests, risks and implications, results, further medical tests and reproductive decisions) reflects the current sociomedical context of patient-centered care and routinized precision medicine. Through shuffling/absorbing, genetic counselors are being situated as uniquely well-suited and integral to prenatal genetic testing, expanding what it means to provide patients medical care in genetics.

The second and third empirical sections delve into two aspects of specialized genomics that are shuffled and absorbed – socioemotionally responsive patient care and the navigation of complex or uncertain patient cases. It illustrates how shuffling/absorbing is made more significant given the current sociomedical context, particularly in how it repositions the counselor from an auxiliary to a more critical provider and uplifts historically feminized professional expertise in the process. The second section shows how the rise in patient advocacy and related patient-centered care (Epstein 1995; Mukherjee 2020; Navon 2019) highlights the need for *socioemotionally responsive healthcare* – a skill that is core to genetic counselors training and expertise (Master's Programs Ethnography 2021). As they absorb and re-medicalize socioemotional tasks, counselors highlight that their approach to prenatal care is attentive to patients' personal needs and integral for patient-centered reproductive care. Subsequently, the third empirical section illuminates how the increasing routinization of genomic medicine in prenatal care ushers *more complexity and ambiguity into patient-provider interactions around genetic testing* – another situation that genetic counselors are expertly trained to manage (Master's Programs Ethnography 2021). Prenatal genetic testing does not always yield clear or conclusive results. Probabilistic results can also be difficult for patients to comprehend, and patients may struggle to understand the diagnostic implications of a particularly variable condition (as discussed in the prior chapter about sex chromosome aneuploidies). As genetic counselors absorb these responsibilities, they emphasize their ability to expertly navigate the often-occurring ambiguity and complexity in genetic testing – once again, allowing them to re-medicalize this specialized expertise as responsive to the current sociomedical moment of routinized prenatal genetic testing. Taken together, genetic counselors are defining their expertise around socioemotionally responsive patient-centered care and navigating ambiguous cases as 'cultural health capital' that is fitting for patients' emerging needs in precision medicine (Shim 2010).

Overall, this chapter shows how expert roles can be cooperatively enhanced and remade given ongoing transformations in biomedicine and sociomedical contexts (Abbott 1983; Eyal 2013; Navon 2019). It illuminates how groups can remake expert roles as they dialectically define their scope of practice in relation to one another, situating these roles in contemporaneous sociocultural, technological, and institutional arrangements. That particular approaches to healthcare expertise can emerge as being more valuable in a given patient care context (transforming expert roles in their image) has important implications for wider social experiences relating to healthcare practices and technologies. In the case of genetic counselors, ongoing transformations in expert roles and expertise are challenging decades of rooted patterns in how 'best practices' and valuable expertise have been defined in genomic medicine, bringing feminized expertise and professional groups to the forefront of the field. As such, genetic counselors' approach to patientcare and increasingly centralized role may both resemble and inform what it means to provide equitable healthcare in reproductive genomic medicine moving forward. These implications are particularly pertinent to reproductive medicine, where prenatal genetic testing is being routinized and can inform decisions including whether a pregnancy is continued, terminated, or provided critical medical interventions.

Shuffling and Strategically Absorbing Specialized Genomics Responsibilities

Reproductive physicians shuffle tasks involving specialized genomic expertise over to genetic counselors, who in turn absorb these responsibilities and frame them definitively within their scope of expertise. This process unfolds as pregnant patients move through the reproductive healthcare

provider workflow. Typically, pregnant patients will first be informed of the opportunity for prenatal genetic testing at their early visits with an obstetrician gynecologist (ObGyn). Depending on the practice, and in most cases wherein ObGyns are constrained for time and referral resources, the ObGyn may offer and order prenatal testing (e.g., NIPT) for the patient. Alternatively, if the physician's practice has the resources and affiliations with a genetic counseling program, they may refer patients to a genetic counselor who goes over various testing options and helps potential parents decide on a testing approach if any. If a pregnancy poses particular concerns after initial genetic screens or ultrasounds, patients may be referred to a MFM doctor (i.e., perinatologist) who specializes in higher risk pregnancies. Unless a patient sees a MFM specialist from the start of their pregnancy, they typically do not consult perinatologists until 12 to 20 weeks into their pregnancy. For those whose pregnancies show concerning genetic findings, genetic counselors would likely be involved in discussing results, further testing options, and alternative reproductive decisions. Based on this workflow, ObGyns are usually the first provider to present patients the opportunity of genetic testing; in cases which require further attention, ongoing support is provided by genetic counselors and perinatologists.

While each provider interacts with patients around prenatal genetic testing, they have differing levels of relevant expertise. Thus, a patient's testing experience is substantially shaped by the particular experts they ultimately interact with. In moving patients through the reproductive care system, physicians carve the boundaries around their professional interactions with patients, determining what is and is not within their desired medical scope when it comes to prenatal genetic testing. While they deem responsibilities regarding pragmatic clinical management of a patient's health needs within their scope, additional responsibilities surrounding in-depth or specialized genomic expertise is de-medicalized and shuffled to genetic counselors, who re-medicalize and readily absorb these tasks into their roles. Through dialectical shuffling/absorbing, genetic counselors gradually become more integral healthcare providers for the genomics healthcare era, as they are willing and able to take on the specialized knowledge and expertise required to thoroughly inform patients about their prenatal testing options and reproductive choices. In this way, their feminized profession and expertise become more centralized in addressing patients' emergent needs as well as expanding the values around providing equitable healthcare in this patient-centered, genomic healthcare context.

Doctors. Before delving into how and why doctors shuffle specialized genomics expertise to genetic counselors, I first explain how physicians define their professional jurisdictions to exclude and de-medicalize in-depth genomics engagement with patients. Both ObGyns and perinatologists (used interchangeably with 'MFM' or maternal fetal medicine specialist) assert that genetics is not their specialty but rather a smaller part of their broader set of responsibilities toward pregnancy management. They frame their jobs as managing the 'big picture' and 'objective' medical needs of a person's pregnancy, with genetics being just one aspect of this. Compared to ObGyns, perinatologists receive more specialized training in genomics, though almost every perinatologist interviewed relied on a genetic counselor (or expressed wanting to, if their practices did not have the resources or established workflow to refer to counselors) when dealing with genetic tests. ObGyns, on the other hand, described prenatal genetic testing as a part of their routine procedures, but noted limited understanding of genomics. An ObGyn at a large private hospital describes:

> "It's that our scope of knowledge is just not there, […] it wouldn't be fair to the patient to try and counsel them about everything just because we don't have all of that knowledge base and we don't have the time. So, my role is to function as the central person for that patient's pregnancy, and if they need the high-risk Ob, then I will refer them; if they need genetics, I will refer them. […]. I'm kind of like their PCP (primary care provider), in a sense, for their pregnancy. […] It's the fundamentals, but beyond that, they need to go towards the specialist. Because (the specialist) just focuses on their own niche and they have longer visits." (Dr. Das Interview 2021)

ObGyns like Dr. Das do not consider themselves to be genetics experts. It is not a part of their established professional jurisdiction. Dr. Das underscores that a having specialty genetic knowledge is not a requirement for his expert medical role; rather, he connects patients to medical resources and manages their clinical needs. Still, although ObGyns admittedly lacked genetics expertise, they offered and often ordered genetic testing for patients – a practice that most genetic counselors took issue with – because the American College of ObGyns recommends its practitioners offer prenatal genetic testing to all pregnant patients and enables doctors to order these tests. In particular, for many ObGyns who did not have sufficient access to genetic counselors at their practices, ordering these genetic tests for patients was standard; only if the results showed any findings or if further diagnostic testing was required would patients be referred to genetic counseling. In general, ObGyns discussed offering NIPT, California Prenatal Screening Program tests, and nuchal translucency (a screening to check for neural tube conditions) to all patients, each considered within the scope of prenatal genetic testing. However, as both ObGyns and counselors described in interviews, ObGyns provided only a brief and often incomplete explanation of these tests before consenting patients and placing relevant orders. For example, ObGyn Dr. Das goes on to describe how he presents prenatal genetic testing via NIPT to pregnant patients at their first clinic encounter:

> "I ask them if they are interested in genetic screening for the baby. And then I go through that, that it screens for trisomy 21, 18, 13, or if they want to know the gender. And most patients will say yes to that and then we just counsel them that it's a blood test that tests the baby's DNA and the moms blood. Most of them say 'yes,' like 80% 85% will say 'yes' to it. And it doesn't really go beyond that." (Dr. Das Interview 2021)

Dr. Das' approach is representative of other ObGyns interviewed, all of whom depicted basic or surface-level counseling when presenting prenatal genetic testing (especially NIPT) to pregnant patients. There was no discussion of the potential drawbacks of NIPT, such as false positives or inconclusive results, preparation around expecting risk- or probability-based results, or counseling around how prospective parents might be able to use the results in informing their family-building or understanding of disability; each of these were components that genetic counselors covered in-depth when presenting patients with NIPT. Thus, doctors' approach to presenting prenatal genetic testing raised issues around patients' informed consent surrounding the types of genetic tests they are opting into and the possibility for concerning results that they may not have been prepared to handle.

There are also time constraints on ObGyns' appointment structures that did not allow them to delve into genetic testing information. ObGyns, including Dr. Das, emphasized that their briefer patient

visits and breadth of responsibilities precluded providing in-depth genomic knowledge. Dr. Das goes on to tell me that he spends "no more than 30 seconds talking about an NIPT" (which is in stark contrast to genetic counselors, who often spend entire 30 minute to 1 hour counseling sessions discussing NIPT; Genetic Counseling Ethnography 2021) because he also needs to cover the patient's medical and pregnancy history, perform an ultrasound, pelvic exam, and breast exam, and discuss their medications during their first clinic meeting. He remarks, "trying to put all that into a 30-minute visit is close to impossible (Dr. Das Interview 2021)."

While MFMs have more specialized knowledge in genomics than ObGyns, they also consider prenatal genetic testing as one piece of their larger expert role, emphasizing their other medically actionable and clinical responsibilities. That is, genetic testing is one component of their broader role in managing high risk pregnancies and must be understood within the context of other medical markers relevant to the patient's pregnancy. Further, although MFMs certainly interact with patients around genetic testing, this often occurs later in a pregnancy. Typically, MFMs discuss secondary or confirmatory diagnostic tests (i.e., amniocentesis, chorionic villus sampling) with patients, to understand genetic findings on initial genetic screens (e.g., NIPT) that have already been conducted under the care of an ObGyn and/or genetic counselor. As such, patients have often been informed about testing, even at a basic level, before meeting with an MFM. Accordingly, one perinatalogist describes that while genetics is an integral part of her daily workflow, it is not necessarily her main medical expertise:

> "Prenatal genetics is part of my every day, but it's definitely not my expertise. I have a pretty superficial background in it […]. Once you get into more of the specifics, then we have genetic counselors that refer patients to. I'm definitely kind of at the surface." (Dr. Paschel Interview 2021)
>
> Another MFM provided more details as to her expertise and professional responsibilities, illustrating how genetics was one a component of this larger clinical role:
>
> "(My role is) a spectrum. I spend most of my time doing ultrasounds and consults […]. And then, I definitely work with the genetic counselors if there's any kind of abnormal case and higher needs, to go over the results. I also work in our [treatment] center, which is like a second opinion referral for different kinds of abnormalities, some of which are genetics some aren't. Some are just kind of a spectrum of abnormalities and pregnancy complications, not all related to genetics." (Dr. Kapoor Interview)

As MFMs Dr. Kapoor and Dr. Paschel define their professional scope, they emphasize that their expert role is to manage higher risk pregnancies as a whole, with genetics being just one aspect of this process. Both Dr. Kapoor and Dr. Paschel also worked at highly resourced research centers with well-established genetics departments, so they could rely on genetic counselors to handle any specialized or in-depth genetic testing cases. For other perinatologists who are working at institutions that are not as well-resourced and lack consistent access to genetic counselors, the approach to prenatal genetic testing involved taking on much of the counseling themselves, unless the patient case was particularly complex. In practices where accessing a genetic counselor was not possible, MFMs presented early testing (i.e., NIPT, nuchal translucency) as an explicit recommendation for all pregnant patients (markedly different from genetic counselors presenting

testing as an optional individual decision; Genetic Counseling Ethnography 2021). In these instances, MFMs took more of a standardized approach to ordering prenatal tests for patients, compared to genetic counselors' tailored and specialized approach with each patient (which I discuss in greater detail later in this section).

While MFMs and ObGyns both take on some degree of genetic counseling when discussing prenatal genetic tests with patients, and define their professional scopes accordingly, these physicians do not take on cases that require specialized or in-depth genomics knowledge or expertise. For cases that were particularly complex, presented with concerning genetic findings, or required further testing and decision-making, physicians shifted (or, 'shuffled') this responsibility to genetic counselors. For example, doctors discuss sending any pregnant patient who has a family history of particular conditions to the genetic counselor:

> "If someone in the family was ever diagnosed with Down Syndrome or an intellectual disability […] I would put in the genetics referral, just because I don't have enough background or enough experience to isolate those as something that does or does not require genetics consult. If there's any question about a family member who had some sort of care, requiring any sort of cognitive defects, I'll just refer them to genetics, and they spend a dedicated 45-minute appointment to just talk about family history. I don't get into it more than if it's a 'yes' or 'no,' then then the genetics referral goes in." (Dr. Das Interview 2021)

> "For a patient who has either a family history or personal history of birth defects, they would need to see genetic counseling. Sometimes things are multifactorial. A baby could be born with some birth defect that may or may not be actually chromosomal or genetic but may have some genetic component. So any patient who has that kind of history, I would recommend that they talk to the counselor." (Dr. Huberman Interview 2021)

Across practices, patients presenting higher risks, showing genetic anomalies on early screens, or requiring further testing would be shuffled over to counselors. In these moments, doctors de-medicalized this level of specialty patient engagement around genomics as outside their expert medical roles, insisting they needed to keep their focus on managing a patient's larger clinical needs and medical interventions. Doctors also relied on genetic counselors to navigate more detailed testing logistics related to costs and insurance coverage, which is critical to how patients experience and access healthcare in the United States. An ObGyn at a large community hospital stated, "in terms of NIPT, I'm very confused all the time about who's eligible, how would it be covered, and so if a patient asks about it or wants it, I just send them to the genetic counselor (Dr. Huberman Interview 2021)." Similarly, a perinatologist noted, "the cost and stuff like insurance coverage is not something that I am very good at. That's another thing that I defer to the genetic counselor to review with (patients) (Dr. Paschel Interview 2021)." These responsibilities were seen as administrative and non-medical, and thus shuffled to genetic counselors accordingly.

When shuffling responsibilities requiring specialized genomics expertise, many physicians also discussed that genetic counselors were better equipped and trained to tackle these situations. Moreover, they framed genetic counselors' expertise as essential for upholding patients' reproductive values: "(Genetic counselors) are my team members. […] They are absolutely central.

I just think informed decision consent and decision-making are very important. […] So, I don't do this in a silo, I work with board certified geneticists and genetic counselors (Dr. Khoury Interview 2021)." Along these lines, some physicians commented on the necessity of genetic counselors to ensure a more equitable patient experience especially given the rapid commercialization and routinization of the NIPT prenatal genetic screening.[57] Below, both Dr. Khoury and Dr. Leighton problematize the commercialization around NIPT and (perhaps inadvertently) frame shuffling specialized genomics expertise as equitable patient healthcare:

> […] The trouble with like things like cell-free DNA screening is that it went into the hands of people who didn't really know understand the tests or what they're doing with these kinds of tests (*referring to the marketing tactics around NIPT from commercial genomics companies targeting non-genetics professionals including ObGyns and patients*). So, I don't think it should just be a given, […] I think (patients) need to go through counseling from someone who can do it in a way that is very, very thorough." (Dr. Khoury Interview 2021, *clarification added*)

> "The NIPT screening companies did a really hard sell in California to say to Obs, 'you can do this right at the get go with your patients.' So, (patients) were no longer were coming to me or our genetic counselors for that counseling. And that's when things changed. People were coming to me and already had their testing done (Dr. Leighton Interview 2021)."

Physicians highlighted that commercialization – and the related issues around inadequate counseling and lack of informed consent – were most salient for low-income and immigrant patients, many of whom had lower levels of education, language barriers, and health literacy. Because doctors tended to present prenatal genetic testing as a direct recommendation, rather than option, patients were regularly having their blood drawn without knowledge that genetic testing was being conducted. Not only was this an issue for those patients who did not desire such information, but it raised further complications when anomalies were identified without the patient's knowledge. Dr. Kapoor comments on the confusion or discontent that NIPT routinization engenders for patients from marginalized backgrounds, many of whom do not realize their blood draws include a sample for the NIPT genetic test:

> "(Patients being unaware that they have consented to NIPT) happens more than you would think. It's really that disparity in health literacy. There's a lot of patients that just go to a clinic and their ObGyn just orders them a lot of tests. There's a language barrier, there's an understanding barrier. They just do what they're told, and they get all the blood work. Then all of a sudden, the results come. There's a lot of blood work, so you don't even know what you're getting. People have no idea; this is just another thing on that list. And only then do they realize this was a genetic test. And they don't have any opinion about it until there's a finding." (Dr. Kapoor Interview 2021)

[57] NIPT is unique among other prenatal genetic tests in that genetic data can be gleaned from a standard blood draw from the pregnant person, allowing non-genetics physicians to order the test, accelerating its widespread implementation. Several physicians problematized this approach to genetic testing, which was driven by commercial interests and not necessarily the level of counseling required for patients to make informed decisions.

Indeed, almost all physicians described ideally wanting to rely on genetic counselors more for their specialized genomics expertise because it would provide a more thorough and informed patient experience. Doctors shuffle patients' specialized genomics needs to counselors not only because of resources and how they define their medical professional roles, with genomics being just one part of their scope (or in the case of ObGyns, outside their expertise), but also because they see genetic counselors as supporting patients broader (non-medical) needs.

Genetic Counselors. In contrast to physicians, genetic counselors defined their scope of expertise as specifically focused on genomics knowledge and related patient engagement. Rather than being one aspect of their professional roles, or a tangential set of expertise, navigating prenatal genetic tests and the counseling surrounding testing and results entirely comprised genetic counselors' expert roles. Further, genetic counselors assert that they have specialized genomics skills that non-genetics physician providers do not. In particular, they highlight their graduate-level training in being able to choose appropriate genetic tests, understand results, and communicate these to patients. These claims to expertise are evident early in the patient care process, as genetic counselors approach prenatal genetic testing with each patient based on the patient's specific situation and reproductive priorities. So, while most counselors still offered patients a standard set of tests (i.e., California Prenatal Serum Screening Program, NIPT, and diagnostic procedures such as CVS or amniocentesis, and carrier screening), they personalized their counseling and explanation of these options based on the patient's family medical background, specific risk factors, and individual reproductive views toward testing. In this way, genetic counselors re-medicalized their specialized expertise, defining their engagement with patients as more tailored to patients' individual reproductive values and thus integral to supporting medical care. Skylar, the head genetic counselor at a large community practice, summarizes this approach below:

> "If someone has a specific family history, for example a really rare genetic condition that's not included in expanded carrier screening, I would offer them testing based on that if it was available […]. My conversation also frequently changes to tailor it to why a person might want to do this or not do this based on what I'm seeing and their personal and their medical history. […] And how I explain it really varies based on what that person's chance to have one of these things in their fetus actually is and how much I think that they would or would not want this information based on conversations I have with them." (Skylar Interview 2021)

Rather than directly recommending testing, counselors align with precision medicine values to personalize the possibility of testing for each patient and augment their role in reproductive decision-making. While many counselors stated that newer testing technologies were applicable across patient populations, they still sought to understand each patient's medical and personal perspectives to specify testing to their needs. In doing so, counselors re-medicalized their more tailored approach to prenatal testing – which incorporated the patient's reproductive priorities and perspectives on genetic testing from the beginning of prenatal care – as an integral aspect of 'equitable' healthcare.

Per counselors, a part of supporting equitable and accessible medical care also involved translating specialized genomic knowledge for patients, a key responsibility counselors absorbed from doctors. They described carefully attending to patients' understanding such that patients,

regardless of their education or health literacy levels, left the session with an adequate grasp on genetic conditions that may be inherited via pregnancy and how prenatal testing could identify some of these. During their graduate training, genetic counselors learn specific strategies to adapt to patients health literacy needs. For example, many genetic counselors relied on analogies to break down complex genomic information for patients with less existing knowledge of genetics. A recurring example was the 'light bulb':

> "You've got a room with two bulbs, right? […] But there might be a time that one bulb is fused. That would be a carrier status, you would be a carrier of a gene that's not working properly. But say, you pass on these bulbs to your children, and you pass on the bulb that's out, and your partner also passes a bulb that's out. Now that child doesn't have any light in that room, and that's when (the condition) progresses in your child." (Shanta Interview 2021)

Thus, for counselors, doing genetic medicine equitably was intertwined with strategically absorbing specialized genomics and related patient engagement responsibilities from physicians, and carrying these out in a manner that could adapt to patients' diverse needs. They absorbed and re-medicalized adaptive counseling strategies as key to supporting precision medicine overall. As they adapted their counseling strategies, genetic counselors paid attention to patients' unique levels of health literacy, sociocultural backgrounds, income levels, religious views, and language proficiencies. They underscored how this expertise was increasingly necessary as prenatal genetic testing (e.g., NIPT) becomes routinely offered, covered by insurances, and often covered by the state. Given the routinization of testing, the patient population includes those of lower income and literacy levels who otherwise may not have been able to afford testing – and who especially need genetic counselors specialized skills. Below, genetic counselor Amy, who spent many years working at a non-profit hospital, describes her approach for patients with varied socioeconomic backgrounds:

> "What I try to elicit from them is what they know already about science and genetics. […] For example, if someone just moved here from Ecuador and has the equivalent of a fifth-grade education, versus a physician who I work with down the hall, those are going to be very different sessions that I'm going to use very different language. […] I have plenty of patients who say 'science wasn't my thing,' so I kind of take a step back and talk about what the gene is, […] how genes tell our bodies how to work how to grow, how to develop, […] about what diseases we might get, how we get half of our genes from mom and half from our dad, which is why we maybe look like our parents and siblings. […] The language that I'll use will be really dependent on what the patient is comfortable with. […] For (people with less scientific background) it might be more helpful to have pictures and show what's happening (to the chromosomes) or avoid using certain bigger words all together if it's going to get too confusing for them." (Amy Interview 2021).

While widening access to prenatal genetic testing is discussed as a critical step for addressing health disparities, experts must be able to provide care that meets the needs of a quickly diversifying patient population. As Amy described, her counseling strategy is adaptive to patients' varying needs, based on their health and science literacy, language barriers, and other socioeconomic differences. She goes on to discuss that this sensitivity to patients' needs remains

critical beyond pre-test counseling, especially when patients receive results that indicate genetic findings that may warrant further reproductive decisions. Along these lines, genetic counselors noted that their dexterity, in terms of efficiently adjusting education and counseling strategies per patient session, was critical for delivering complex information in a simplified yet comprehensive manner. They discussed (and re-medicalized) their proficiency with specialty genomic knowledge as a boon for equitable patient care, as ensuring patients of all backgrounds appropriately understood their options during pregnancy was essential for uplifting reproductive choice.

While doctors de-medicalized responsibilities around insurance coverage and testing logistics around testing as administrative and tangential to medical work, genetic counselors absorbed and re-medicalized these tasks as a part of their specialized genomics expertise. Counselors were well-informed about insurance coverage for prenatal genetic testing and could navigate tests and results based on various genomics laboratory polices. In contrast to physicians who did not have this in-depth knowledge about testing logistics (and who specifically referred these issues to counselors), genetic counselors were thoughtful in ordering tests that would not financially burden patients, especially those who are low-income. In this way, they re-medicalized these so-called administrative as central to patients' medical care needs, fluidly adapting to patient's personal contexts during a session.

As genetic counselors position their willingness to absorb specialized genomics expertise as within their professional scope and critical for equitable patientcare, they also problematize doctors' approach as harmful to patients' reproductive experiences. In this way, they define strategically absorbing specialized genomics tasks from doctors as critical to ameliorating current gaps in patientcare. Comparing their expertise to that of physicians, counselors critiqued physicians' (mainly ObGyns) approach to recommending and routinely ordering genetic tests without providing adequate in-depth patient education. While counselors understood the various constraints ObGyns faced (e.g., shorter patient consultations, more information to cover), they repeatedly noted that patients often left ObGyn consultations uninformed about what prenatal genetic testing implied, and at times were unaware that they had undergone a genetic test. For most, this inadequate genetic counseling raised red flags around patients' informed consent due to insufficient genomic education, which compromised patients' reproductive choices with regard to testing and their pregnancy experience. Here, genetic counselor Rosie describes their experiences with patients who had received inadequate pre-test counseling and testing from an ObGyn:

> "So many patients are not able to articulate their understanding of this testing […]. Where I get more concerned is around informed consent because we see the paperwork that patients fill out, sign, and date, supposedly indicating whoever ordered testing went over that information with them, and I don't necessarily believe that all of that is happening. […] I feel unsettled because I just don't know to what extent it's been an informed decision or whether it's really just the Ob saying you need this testing and I'm going to order it for you. (Rosie Interview 2021)

Rosie emphasized her concerns around informed consent for patients who had only received prenatal testing counseling from ObGyns, whose expertise did not encompass in-depth genomic knowledge. Other counselors described that ObGyns often ordered prenatal genetic tests without fully understanding what conditions were being screened for or inaccurately interpreted results.

And while MFM doctors had more specialized genomics expertise than ObGyns, their wider scope of their responsibilities often meant patients did not receive enough education specifically about genetics. As such, where genetic counselors underscored optionality in their counseling approach, explicitly providing patients the opportunity to decline all testing (often times, being the first provider to do so), they raised concerns around doctors directing patients to pursue genetic testing as a standardized part of clinical management. Counselors Maggie and Tanya illustrate this below:

> "Doctors almost always tend to be directive. A doctor sees a pregnancy that has a heart defect, and they want the patient to do an amnio[centesis] so they potentially could find out a reason. […] But just because (providers) want that information doesn't mean that's appropriate for the patient. […] I don't understand how hard it is for doctors to understand that if I have an hour set aside to talk to a patient, how do you think you're supposed to get that done in 15 minutes?" (Maggie Interview 2021)

> "Obs (ObGyns) will say, 'we need to know if this baby has this condition,' so we sometimes have patients who didn't want that information but felt pressured by their Obs to (test). […] We occasionally get angry phone calls from the Ob saying, 'why did you tell her she shouldn't have to do this test?' but we have to stand up for (the patient's) decision (Tanya Interview 2021)."

This 'directiveness' of doctors' standardized approach was a major concern across genetic counselors interviewed, as it resonated with issues stemming from genetics' troubled history with eugenics. Across counselors, doctors' directiveness justified why they *needed* to absorb specialized genomics responsibilities from physicians, to address current failings in how prenatal testing was being routinely carried out. Above, both counselors Maggie and Tanya describe that while prenatal genetic testing may be clinically insightful to doctors, it is not always the most appropriate decision given patients individual reproductive values and priorities. They both emphasize the role of the genetic counselor in ensuring equitable care that reflects each patient's choices, illustrating how they not only engage patients thoroughly but also push back on doctors imposing testing on patients. In these cases, counselors mobilize specialized genomics expertise to bolster their professional claims while also expanding what defines equitable patientcare in precision medicine.

Notably, there are tangible consequences for patients who do not receive specialized genomics attention, and when directiveness from physicians becomes the standard approach. Many counselors recounted experiences where doctors ordered the 'wrong' test for pregnant patients because they lacked genetics expertise. A test may be inappropriate or incorrect if patient's medical history, personal preferences, or pregnancy specificities do not align with selections for the type of test, specific conditions tested, or laboratory practices. Errors in ordering tests or neglecting to thoroughly educate parents about the implications of genetic findings can bear harmful consequences. In addition to unnecessary financial losses, such situations can tarnish patients' experience of their pregnancy. In other instances, the outcomes can be more severe, wherein patients terminate pregnancies based on inaccurate information:

> "I think any genetic counselor who works in prenatal (care) will tell you stories about people who get back abnormal NIPT results, and because they're not properly counseled

> about the fact that it's not always a one-to-one correlation, they will terminate a pregnancy based on it. Because no one told them […]. Or, you have someone who gets a carrier screening results, and they're improperly interpreted, […] and they're told 'you can't try to have kids on your own' […] but really, it's just a vitamin deficiency. […]. And the main reason for all this is because the Obs (ObGyns) are ordering the tests." (Mara Interview 2021)

Almost all genetic counselors provided similar examples of cases where patients suffered and made inaccurately informed reproductive decisions due to providers' inadequate genomic expertise. Some described counseling patients who had been told to terminate their pregnancies by physicians: "I'll have people that will say, 'well, my doctor is telling me I should just terminate, that this is really bad, that I have a 25% risk to have a baby with this condition.' Then, I'll find out the baby's dad hasn't even been tested yet (Rosemary Interview 2021)." In these moments, genetic counselors described their roles as attempting to undo some of these harms: "We end up picking up a lot of pieces and doing a lot of reeducation for patients. It becomes damage control (Rosemary Interview 2021)." As counselors noted, 'damage control' involves using their specialized genomics expertise to re-empower patients in making reproductive decisions and moving forward in managing potential findings. In this way, genetic counselors re-medicalized their specialized genomics responsibilities as integral to supporting patients' medical experience. As they absorb these shuffled 'burdens,' counselors prioritize carrying out related patientcare and engagement in a way that is not only fitted to their level of training and focused expertise but also responsive to patient needs and reflective of equitable healthcare as prenatal genetic testing becomes increasingly routinized.

Finally, for counselors, efforts around the NSGC Act further illuminated their arguments around claims to expertise. The Act seeks to appropriately recognize counselors' expertise as a matter of equitable patientcare, in that it would facilitate access to providers who can provide in-depth genomics engagement needed for the shift to precision medicine. In prenatal care, genetic counselors situated the NSGC Act as a way forward in mitigating the harmful consequences that arise when patients receive little to no counseling around prenatal testing, are provided misinformed guidance, or do not undergo a thorough informed consent procedure. Here, counselors saw physicians' lack of support for their Act as unjust gatekeeping that fundamentally contradicted patientcare standards. In Skylar's perspective (which several other genetic counselors shared) much of the resistance to affording counselors' greater professional capacity relates to power struggles rather than patients' best interests:

> "I think the (American Medical Association) is an extremely powerful entity. (These) very powerful organizations […] want physicians to be involved with every step of medical care and the United States. […] It's an understandable although misguided desire to protect the jobs and protect the role, and the power behind being a physician, because there is more than sufficient literature to support the fact that a genetic counselor is the most appropriate person to select genetic testing in a medical context." (Skylar Interview 2021)

Along these lines, many genetic counselors underscored that, unlike ObGyns, they had been specifically trained in genomics with a thorough understanding of testing technologies. As such, they emphasized that patient engagement around genetic testing was well within their professional

scope, and moreover reflected the specialized expertise they developed through their graduate-level education in genetic counseling. Along with their education, counselors underscored that being Board Certified bolstered their expertise and ability to order tests. Accordingly, they positioned their expert roles as more focused and distinct from those of ObGyns, justifying their claim to greater professional autonomy in terms of ordering tests:

> "Just because they're an 'MD,' doesn't mean that they know the right questions to ask the patient, it doesn't mean that they understand the differences between a (variant of uncertain significance) and the pathogenic variant, it doesn't mean that they've been taught any of this stuff. And we spend two years in school, completely focused on that. The rest of our career is focused on that! So, for ordering genetic tests, most genetic counselors are more qualified than most non-genetic doctors to do these things." (Tanya Interview 2021)

Indeed, genetic counselors have already absorbed the task of ordering tests in many ways. In most cases, counselors were effectively ordering tests for patients, but for the barrier of being required to list and bill for this under a physician's name. Both physicians geneticists (e.g., MFMs) and genetic counselors described that while the physician signing off on the genetic test may review the counselor's order, they typically do not provide much oversight as to which tests are most appropriate, acknowledging that this is the counselor's specialty. As such, counselors asserted that they should be recognized for the expertise they are already demonstrating in patient care:

> "Many tests are, in reality, basically ordered by genetic counselors. […] Doctors' part of the care team, in practice, is often just a button click that is changing who the order is under. […] At my former clinic, […] I was always the one who 'ordered' the test, but it was always ordered under their name. So, for insurance the doctor ordered the test, but I always decided in the end which was the test to do […]. Some genetic counselors I know order under the name of a doctor who they basically don't even know or who doesn't really see the patients." (Martha Interview 2021).

While genetic counselors establish that, in practice, they can order tests, they emphasize the significance of the Act in legitimizing this professional autonomy for their expert roles. There was a sense that the expectations of their roles were mismatched with what insurance companies were recognizing and compensating them for, with the Act aimed at ameliorating this inconsistency. Although counselors helped patients and doctors select appropriate prenatal genetic tests, and spent lengthy counseling sessions explaining the complex implications and decision-making options around results, they were not being recognized as an expert representing specialized knowledge within this very scope of practice. There was also frustration that they were being made to operate in a way that did not align with how physician associations limited their scope of practice, which influenced insurance policies; they were being asked to 'absorb' more responsibilities based on their expertise than they were being acknowledged for (i.e., physicians relying on counselors to order prenatal genetic tests, but listing the order under a doctor's name). Many counselors described feeling "insulted" by this: "I talked to genetic counselors who still feel like they're treated like an assistant or an administrative role […] rather than a member of the team, who has specific expertise as well (Rosie Interview 2021)." Unsurprisingly, a critical part of this expertise recognition involves being able to officially order tests and bill for their services with appropriate compensation, as for any other provider.

As doctors shuffle specialized genomics expertise and related in-depth patient engagement to genetic counselors, and often recognize that counselors' are better suited to take on this role, genetic counselors strategically absorb these responsibilities. Unlike doctors who de-medicalize specialized genomics responsibilities as tangential to their medical roles (i.e., ObGyns) or as a smaller part of their larger clinical duties (i.e., MFMs), genetic counselors situate these tasks squarely within their expertise, comprising their entire scope of practice. As such, counselors are willing and able to strategically absorb specialized genomics responsibilities from physicians. Moreover, they re-medicalize their willingness and ability to do so as critical for equitable patientcare in the current sociomedical context. Taken together, genetic counselors see their strategic absorption of specialized genomics responsibilities, and how these efforts are reflected in the Act, as integral to improving equitable access for patients in a landscape where genetic testing is increasingly routinized.

With an understanding of how shuffling/absorbing of specialized genomics and related prenatal patient engagement unfolds, the subsequent sections illustrate two examples of how this process is elevated by ongoing changes in biomedicine. I first turn to how genetic counselors provide counseling that is far more socioemotionally responsive to patients' needs than physicians (with physicians relying on counselors to do so). Importantly, counselors' absorption of this aspect of specialized genomics expertise is medically relevant given the sustained movement around providing patient-centered care. Next, I discuss how doctors rely on genetic counselors to navigate patient engagement in particularly ambiguous or complex genetic testing cases. Once again, this aspect of counselors' absorbed responsibilities is made medically significant given the sociomedical context of increasingly routinized genetic testing, wherein ambiguity and complexity is not only commonplace but inherent to genetic testing technologies. Throughout, we also see how counselors' traditionally feminized expertise are being uplifted. That counselors' expert roles are being more centralized in reproductive medicine is particularly significant, as this presents the possibility for a historically feminized profession to not only gain greater recognition in a largely patriarchal field but also have the potential to define how equitable patientcare should be carried out in future practice.

The Labor of Socioemotionally Responsive Counseling

Providing support that responds to patients' sociocultural contexts and emotional needs is important to genetic testing, particularly given the sustained emphasis on patient-centered care and patient advocacy (Epstein et al. 2005; Navon 2019). This sensitivity toward patients' broader personal circumstances is especially essential after they receive a diagnosis or test finding, potentially suggesting their child may be born with a genetic condition or disability. During these sessions, patients often seek support to make difficult decisions, including how they might approach continuing or terminating their pregnancy, or what medical interventions their future child may need. Although both physicians and counselors understand the importance of socioemotionally responsive clinical care, genetic counselors consider this a critical aspect of their expertise while doctors tend to de-medicalize and push it outside their professional jurisdictions. As counselors absorb this responsibility from doctors, they re-medicalize their socioemotionally responsive approach to genomic medicine as particularly valuable and necessary given today's patient-centered care setting. Given the history around genetic counseling as a feminized profession, whose expertise have been less valued compared to other experts in the medical field,

uplifting counselors' roles based on their socioemotionally responsive counseling is noteworthy as it revalorizes traditionally feminized skills in the face of emerging healthcare needs. Given that labor involving socioemotional engagement is often considered 'care work' and underrecognized for its efforts, this transformation in the genetic counselor's role broaches an opportunity to shift longstanding patterns in how knowledge and expertise are produced and valued in medicine.

Doctors. Doctors do not consider socioemotionally responsive counseling an aspect of their expert medical role and often refer emotionally complex cases to genetic counselors. Some of this workflow can be attributed to doctors having shorter sessions with patients, compared to genetic counselors who have longer consultations to delve into patients personal contexts. However, physicians also define their expert role as providing "objective" or "rational" clinical care and interventions to patients, with socioemotionally responsive counseling lying outside what constitutes medical work. As such, doctors typically engage with patients to carry out necessary medical care *after* patients have received support from a counselor and made potentially difficult decisions regarding their pregnancy. Where genetic counselors pair their genomic knowledge and socioemotional counseling expertise when interacting with patients, doctors focus their role on the logistics necessary for clinical care, not explicitly considering emotional management as an aspect of this. Dr. Das, an ObGyn at a large hospital, describes how he approaches patients who have received a prenatal diagnosis or potentially concerning results, noting how he readily refers them to genetic counselors:

> I pretty much talk about trisomy 21 being Down Syndrome and say (trisomy) 18 and 13 are a little bit more severe and may not be compatible with life. […] That's pretty much the extent of my conversation (about the diagnosis). […] It's more of a supportive role at that point. Then, I just tell them, 'We have (genetic counselors) who will discuss this (result) and counsel you more on what this means for your pregnancy […]. If this pregnancy is desired or undesired, that will be a conversation we can have after you speak with genetics.' After (they see genetics), I can counsel them, if they want to terminate, on what do we do then. (Dr. Das Interview 2021)

If a patient's genetic test comes back with possible findings, doctors almost immediately refer the patient to genetic counselors. Although many physicians describe ideally wanting to address patient's socioemotional needs more, ultimately counselors took on these cases requiring more complex decision-making based on results. As Dr. Das illustrates, he refers patients with concerning test results to genetic counselors, who take on this in-depth and socioemotionally involved patient engagement. When he sees the patient again, they have already received genetic counseling and have possibly made emotionally challenging reproductive decisions alongside their counselor. At this point, the physician focuses on explaining and carrying out any medical interventions (e.g., termination procedures). In this way, doctors relied on counselors to build a deeper relationship with the patient; they expected counselors to provide patients in-depth support regarding implications findings may have for their child to-be, necessary follow-ups, potential disabilities and medical interventions, disability education, and referrals to relevant resources. Doctors repeatedly highlighted that genetic counselors could be more "involved" with patients, enabling these socioemotionally responsive conversations, which then facilitated doctors' 'objective' role centered on treatments and interventions. Here, we see how doctors shuffle in-

depth genomics responsibilities they consider peripheral to practicing medicine, even though these tasks (that counselors absorb and re-medicalize) are crucial to patients' medical care overall.

When supporting patients through difficult test findings or complex decisions, doctors aimed to outline subsequent clinical steps. These could include gathering information from other specialists about expectations related to the genetic condition, presenting additional testing options to better understand the condition, preparing for potential medical interventions, and outlining the clinical processes that go into termination alongside its legal timeframe. At times, physicians also provided medical recommendations when discussing possible next steps, describing how this provided the patient with useful future directions to focus on. If, for example, a patient's genetic screen indicated probability of a particular condition, many doctors recommended that patients conduct a diagnostic test to confirm findings or learn more about the genetic etiology before making pregnancy decisions. They described these recommendations as helping patients establish expectations of various outcomes, or plan for complex situations like resuscitating a baby with a life threating condition. However, in many cases, these recommendations were also made to aid the doctor's ability to clinically manage the pregnancy. For example, Dr. Khoury, an MFM who handles particularly complex pregnancies, describes her approach with patients who were firm on continuing their pregnancy with genetic conditions:

> I still strongly recommend genetic testing so we can better take care of the baby. So, if it is a specific cardiac lesion, if I know it's a DiGeorge cardiac lesion versus just that cardiac lesion, I know that they're going to have issues with other organs. I'm going to be aware of things, our NICU (neonatal intensive care unit) team is going to be ready. We'll know if we'll offer or not offer surgery based on outcome. So, it helps to optimize care for the neonate at delivery." (Dr. Khoury Interview 2021)

Here, doctors medicalize their scope as focusing on 'next steps' a patient should consider that would facilitate objective needs for clinical care. They described presenting medical information to patients as "layers" or "chapters" based on the available medical options, the first of which concerned clinical steps that needed to happen before the legal limit on termination (in California, under 24 weeks or until the fetus weighs 500 grams, though it can be subjective based on the pregnancy). This included allowing patients to plan for further testing and wait periods between results, as well as enough emotional space to consider results and reproductive decisions. After the termination window, doctors only presented patients with clinical decisions surrounding the child's delivery, such as preparing for the baby's demise or consulting with specialists about potential surgeries upon birth. Importantly, doctors did not discuss their medical roles as centered on providing the socioemotional support that patients often require in situations where their child to-be may have genetic health complications. Instead, they often relied on counselors to explain to patients why certain recommendations may benefit their reproductive experience. For their part, genetic counselors mediated between patients and physicians, tackling the emotional labor needed to explain how a doctor's medical recommendations may be beneficial and help patients make decisions aligned with their reproductive values. As such, we see the complementary roles these experts hold in providing medical care, with counselors' socioemotional engagement expanding doctors 'objective' approach to clinical interventions.

Overall, while physicians' noted time constraints prevented more socioemotional engagement with patients, they also saw their roles as being distinct from this type of patient interaction. Several doctors defined their expert role as providing "objective" medical information and carrying out evidence-based interventions. More emotionally involved patient engagement was shuffled to genetic counselors, as physicians fundamentally de-medicalized this as outside their scope of practice. Dr. Khoury stated: "My role is to provide (patients) the best evidence, the objective data, to make a decision that they can live with. […]. So, if someone has trisomy 18 and they say, 'what should I do,' I say, 'make a decision that's right for you, but if we do nothing the baby will die on average at these many days.' […] I don't sugarcoat it […]. (Dr. Khoury Interview 2021). In defining their scope, many doctors emphasized that they primarily dealt with 'medical facts,' and at times, emotional engagement was considered interfering with this 'objective' or 'rational' role. As such, physicians delivered information differently from genetic counselors; where genetic counselors paired genetic information with a patient's broader context, doctors prioritized a more straightforward approach, shuffling patient engagement necessitating more attention to socioemotional needs to counselors. Further, while doctors valued genetic counselors' deeper relationships with patients, they saw these socioemotionally involved conversations as going beyond the parameters of practicing medicine as a physician. Dr. Kapoor, an MFM at an academic medical institution, explains:

> "There's a line. If I think there's a maternal risk event, I'm very clear (about recommended interventions). And there are some situations when continuing the pregnancy with a certain kind of abnormality definitely poses some risk. In those cases, I can be a lot more directive, but there's some cases where I cannot. […] We have counseling services for (patients) to think about things in different ways […]. When things like this happen, it often shows you a lot about people's relationships with their partners and unearths so much that it's actually way beyond the scope of what you can do as practicing medicine." (Dr. Kapoor Interview 2021).

Doctors' definition of 'medical practice' limits their emotional labor to only that which is necessary for clinical interventions; genetic counselors, on the other hand, become involved in this labor (which is a part of the in-depth patient engagement they provide), at times going as far as counseling partners and families together. Moreover, doctors rely on counselors to take on this more socioemotionally engaged role, framing it as the genetic counselor's "responsibility" rather than their own. And while some doctors uplifted genetic counselors' specialized counseling skills when it came to socioemotionally involved patient engagement, others emphasized that counseling was not particularly worthwhile for a physician. Dr. Liang, a recently retired ObGyn, described: "(Genetic counseling) is a lot, it's kind of repetitious, and it's a service […] worthwhile as a teacher. But it's certainly not intellectually stimulating for you as a physician to have to go through those things. It's kind of rote work, just the bread and butter (Dr. Liang Interview 2021)." De-medicalizing counseling services as education rather than clinical management, Dr. Liang comments broadly on what expertise counts as 'medical.' Other doctors placed more value on counseling expertise in patientcare, but similarly did not find it to be appealing to their medical practice as physicians. As such, doctors justified shuffling this aspect of genomic medicine to genetic counselors, in part because it did not align with their definition of a physician's expert medical role.

Genetic Counselors. As doctors shuffle socioemotional patientcare, genetic counselors absorbed these responsibilities within their scope and specialized training, re-medicalizing this expertise as critical to delivering patient-centered genomic medicine. They conceived of their expert role as fundamentally incorporating this specialized skill when providing genomics-based healthcare. Thus, emotional labor, though largely unattended to by doctors (and shuffled accordingly), was critical to how genetic counselors interpret genetic tests and center patients' reproductive values. When prenatal genetic tests indicate a fetus may have a genetic condition or disability, counselors convey this to patients and walk them through various reproductive options, providing counseling that is not only informative but empathetic. These sessions can be especially complex from an emotional standpoint, as some patients learn that their fetus may have a challenging disability or life-threating condition such as Edward (trisomy 13) or Patau Syndrome (trisomy 18), both of which are screened for using NIPT. As one genetic counselor notes, "when I'm telling you your child probably has trisomy 13, and with your pregnancy will likely pass away, emotion cannot be completely removed (Maggie Interview 2021)." Accordingly, counselors are experts in patient-centered emotional management – a core responsibility they absorb from physicians – and are integral to supporting patients' medical needs. Below, Skylar describes how she addresses patients' socioemotional needs when it comes to follow-up diagnostic genetic testing (typically after initial screens like NIPT indicate possibility of a genetic condition in the fetus):

> One of the things that I ask people before they do a diagnostic procedure (for Down Syndrome) is what they think they would do with this information. There are some people who are very clear that they would terminate. And some people who are extremely clear that they do not want to hear about termination. […] And there are some people who say, 'I don't know.' So, when I get the diagnosis back, we've already gone over at least some of the features that we would expect before even doing the test. […] That context makes it easier in the sense that I am much more neutral when I talk about the diagnosis with someone who might make a pregnancy decision. And I'm just less so with someone who says I never want to hear you talk about termination again […]. (Skylar Interview 2021)

That their approach to patient engagement already pairs socioemotional responsiveness with delivering in-depth genomic education positions counselors to readily absorb patient responsibilities around emotional management from physicians. For patients who receive difficult diagnoses (or possible genetic findings), genetic counselors implement strategies to support them in understanding the results medically as well as process the implications for their pregnancy from an emotional standpoint. Here, Skylar's approach to ordering testing and counseling is structured primarily by patients' reproductive values and socioemotional needs, varying based on what each expecting parent feels is right for their family. Thus, the way that counselors deliver genomic medicine is not considered separate from attending to patients' socioemotional needs, grounding how they help prepare patients and manage their testing experience accordingly. As prenatal findings after genetic testing become more commonplace with advanced technologies, counselors underscore this expertise in socioemotional engagement as medically necessary and valuable in a growing patient-centered care landscape.

Genetic counselors' socioemotionally responsive approach is especially relevant when patients contend with difficult decisions regarding the pregnancy. In these instances, counselors adapted the way they presented tests, discussed expectations, and eventually presented results to patients

based on each of their personal circumstances. Throughout, the patient is at the center of the testing process; genetic counselors ensured that any medical interventions that may follow a prenatal finding were structured around patients' values, needs, and desires, as opposed to prescribed or recommended by a provider. In absorbing socioemotionally responsive patientcare, genetic counselors become the key providers responsible for guiding patients through reproductive decision-making. As genetic counselor Betty describes, the reproductive counseling process is often more nuanced than a choice to terminate or continue:

> "(Patients) might have to choose between induction versus a D&E (dilation and evacuation), which might involve an injection to stop the heartbeat. There are all these nuances […]. Do you want handprints? Fingerprints? A memory box? We get them started on those thoughts to consider what they need to honor this pregnancy. Some people […] don't want to see the ultrasound, don't want ashes, don't want any memories, don't want to be awake for the procedure, don't want to induce. They have a different way that they're going to cope with this. But some people want to have that induction, they want to hold their baby until the baby passes away. I feel so much for them […]. One time, a woman had asked for her infant to be able to feel the sun on their skin before they passed away. We'll try anything to help them with that process, so it feels as comfortable and as meaningful to them." (Betty Interview 2021)

As seen in Betty's account, counselors perform intensive and essential emotional labor, often empathizing deeply with their patients. Other counselors described patients asking for a birth certificate so that they can name their child or opting to deliver by induction to experience birthing labor. And while many patients did not seek this approach to termination, for others these options provided important closure and attention to their emotional situation. Though the medical procedures for pregnancy termination may be clinically standardized – the 'objective' aspects which physicians tend to focus on – the medicalized process by which a patient experiences this, and the various decisions they can make to cope with their loss, is far more complex and necessitates adequate expertise. In this context, genetic counselors' focus on ensuring patients receive not only appropriate medical attention, but moreover interventions that speak to their socioemotional needs and personal contexts is critical for delivering genomic medicine in way that values and reflects patient-centered care.

Here, it is also worth noting that this type of emotional 'feminized' labor has long been undermined and undervalued, especially in the medical field. Mobilizing for greater recognition of their socioemotional expertise represents a key issue that counselors are tackling via their NSGC Act, where they position their socioemotionally responsive approach as critical to patient medical needs and wellbeing (Higgins 2021; National Society of Genetic Counselors 2021b, 2022). This makes it all the more significant that genetic counselors are absorbing and re-medicalizing these responsibilities from physicians, as it possibly paves the way to transform how traditionally feminized knowledge is valued and legitimized. If the Act is successful, it would afford counselors a louder voice in defining patient-centered genomic medicine, elevating and revalorizing emotional labor in the process.

For those who decide to continue pregnancies with genetic conditions, or who seek more support, genetic counselors are a central point of information and mediator connecting them to other

resources. They are often the first provider who discuss genetic conditions and disability implications in-depth with patients, providing the resources patients need to make informed decisions that reflect their reproductive values. In these conversations, counselors emphasize remaining neutral while empathizing with patients' perspectives, which can also be more challenging when patients face language or health literacy barriers. A common approach counselors took was to explain the possible medical needs related to a particular genetic condition while noting that disabilities may present in a range of ways that prenatal testing cannot readily reveal. It was also very common for genetic counselors to refer patients to other specialists who could provide additional perspectives on what their future child's medical needs may be. For example, counselors suggested patients speak with a pediatric cardiologist if their fetus showed heart concerns, or a behavioral therapist if the diagnosed condition indicated cognitive needs. These additional resources were essential to helping patients come to terms with their reproductive decisions in ways that prioritized their personal and emotional contexts. The following patient example that counselor Rosemary illustrates how genetic counselors take on critical socioemotionally responsive counseling as a central part of patients' medical care:

> "There was a case where the baby had anencephaly. And (the mother) was very Catholic. She thought she wanted to interrupt the pregnancy, but she couldn't wrap her head around it. She kept saying, 'I'm killing my baby, it's against my religion, I can't do this.' […] And there used to be a priest at [medical center] and he was also trained as a genetic counselor. He would talk to patients who were very religious, very Catholic, and with issues like this person had. I was able to put her in touch with him, which was so helpful for her. In the end, she ended up electing to interrupt the pregnancy." (Rosemary Interview 2021)

Anencephaly has no cure. It is a neural tube condition wherein a fetus is missing parts of its brain and skull. If a baby is born with anencephaly, it is typically stillborn or may survive up to a few days. Rosemary illustrates how important counselors are to helping patients cope with reproductive decisions that may be difficult given their personal situations or backgrounds. While termination is a common decision in anencephaly cases, for a patient who struggles with the concept of abortion, being able to make sense of their decision in the context of their religion can be a core component of their medical management. In these instances, we see how genetic counselors re-medicalize socioemotionally responsive care, positioning this as not only core to their specialized genomic expertise but also necessary for patient-centered healthcare.

Prenatal genetic testing can also 'spillover' into patients' partners, parents, children, and others in their close network. Multiple people may weigh in on reproductive decisions or findings may reveal health implications for other biological family members (Timmermans and Buchbinder 2012; Timmermans and Stivers 2017). Often, doctors define the additional socioemotional labor involving family members outside their professional scope (recall Dr. Kapoor, the MFM who suggested this type of patient engagement was "way beyond the scope of what you can do as practicing medicine"). As doctors shuffle these responsibilities, genetic counselors absorb them as a part of their socioemotionally informed approach to genetic testing. In doing so, they become crucial medical mediators as they support patient units (e.g., families, couples) as a whole. It was common for patients to bring their partners, children, or parents to consultations, so that counselors could provide support for the family together (Genetic Counseling Ethnography 2021). Genetic

counselor Kim discusses how influential a pregnant patient's partner can be, in particular, and notes how she attends to this important aspect of patient's socioemotional needs:

> "The partner's influence is big. I have this one patient whose baby on the ultrasound has really severe abnormalities. It's not moving very much, and it's clear that there's something wrong. She wanted to terminate but she wanted to talk to her husband. She said her husband doesn't want to terminate as it's his first child. […] She wanted me to talk to the husband, which I did. Then, they ultimately decided together that they would continue the pregnancy." (Kim Interview 2021)

Recognizing the broader implications of prenatal genetic data, including providing counseling others in the patient's circle who may be impacted by findings, was key to genetic counselors' approach to patient-centered and socioemotionally informed genomic medicine. As patients often included their loved ones in their reproductive decision-making, it was especially important that genetic counselors could create a sensitive and emotionally informed space for collective discussion and questions during this process (while still centering the pregnant patient's needs). Indeed, in the patient consultations I observed, there were numerous instances where patients brought family members to speak with genetic counselors during particularly difficult sessions. In one case, a pregnant young woman with autism arrived at a counseling session with her sister, and the counselor had to inform the patient that her fetus had Fragile X Syndrome.[58] This was a particularly challenging case, as the counselor needed to centralize the socioemotional context of patient unit (pregnant person and her sister) as a whole, while trying to ensure that the pregnant patient was informed of her situation and consented to reproductive decisions reflecting primarily her values (rather than her sister's, who was vocally opposed to termination or in-utero interventions). In other instances, pregnant patients brought their partners or their parents to consultations where they received difficult news about their fetus' genetic conditions, with many breaking down in tears during these sessions. Genetic counselors not only had to navigate these emotionally heavy consultations with sensitivity and empathy, but they had to continually deliver the relevant medical genetic information in a manner that respected each personal context. The socioemotional labor that counselors absorbed, and encompassed within their approach to genomics carre, was thus critical to emerging patient-centered care needs in prenatal genetic medicine. (Genetic Counseling Ethnography 2021)

Finally, it is important to emphasize that genetic counselors did not consider this socioemotionally responsive approach to counseling as an additional or supplementary aspect of their expertise, but rather part-and-parcel with how they defined their scope of practice within genomic medicine. There was no counseling that was not socioemotionally responsive. As such, genetic counselors not only had the expert capacity to take on this shuffled 'burden' from doctors, but they were able to re-medicalize it within their extant skills that prioritized patient-centered care. Unlike many physicians, genetic counselors underscored that they are distinctly trained to deliver

[58] Fragile X Syndrome is described by the Centers for Disease Control and Prevention as "rare genetic disorder that has a major effect on a person's life." While it is one of the most common genetic causes of intellectual and developmental disabilities, there are still many unknowns about how this syndrome presents (Navon 2021). The phenotypical presentations of Fragile X Syndrome can vary greatly; however, affected individuals are described as having "problems with memory, abstract thinking, problem solving, and planning" as well as "social and behavioral problems, difficulty with learning, developmental delays, and autism spectrum disorder." (Centers for Disease Control and Prevention 2022)

socioemotionally responsive counseling using intentional strategies and selective language. Compared to physicians, counselors were also more sensitive when delivering results and described the harmful consequences they had to mitigate – what they termed, 'damage control' – when physicians took other approaches. Several counselors described times when doctors or their office staff delivered results to patients over the phone, inaccurately interpreting results or presenting findings as definitive health outcomes rather than odds-based information. In these cases, patients would be left unnecessarily scared and misunderstood the implications for their pregnancy, which counselors had to ameliorate. In these moments, genetic counselors emphasized the applicability of their specific socioemotionally responsive training, as Maggie, a senior counselor at a large private hospital, explains:

> As a profession, we really employ a lot of the typical counseling tools, whether it's active listening, or mirroring, or asking the uncomfortable questions, […] playing devil's advocate […]. These are things that we build on in our career and that we're trained to do. If you hear a medical provider, an MFM for example, talking about (genetic results) they don't have as much of that training […]. But we really do have specialized training to do that. We learn it in school. We try to use words that don't have as much connotation to them, that aren't so heavily laden with background. […] Like 'risk' just has that connotation of bad versus 'chance,' which is a more neutral term. I'm talking about the same thing, but people don't tend to have as much a emotional reaction to the word chance versus risk. (Maggie Interview 2021)

Counselors' affective emotional labor did not only derive from empathy but was a specific set of skills they developed to become experts, fundamentally structuring their understanding of prenatal genetic technologies and related medical interventions. As such, socioemotionally informed counseling is integral to genetic counselor's approach to providing genomics-related patientcare; it is fundamentally a part of their defined expertise in this field, rather than an additional 'burden' or responsibility as it was often framed by physicians.

On par with their specialized ability to accurately interpret genetic tests and results, counselors' socioemotional expertise is critical to supporting patient families as they undergo genetic prenatal testing. Thus, as doctors de-medicalize and shuffle socioemotional patient engagement, genetic counselors strategically absorb and re-medicalize this expertise as critical to supporting patient needs in precision medicine. For counselors, being able to harness their specialized genomic knowledge in a way that made sense for patients, both in terms of medical education and emotional needs, was a crucial part of delivering equitable healthcare in a patient-centered context. The backdrop of the sustained patient-centered care movement further elevates counselors' expert roles and approach to genomic medicine, revalorizing their 'feminized' expertise around socioemotional labor as fundamentally necessary for addressing patients' needs as genetic testing becomes commonplace in reproductive practices.

Navigating Ambiguity and Complex Genetic Testing Cases

Uncertainty is inherent to prenatal genetic testing. Genetic screenings (e.g., NIPT) results provide probability-based indications of a child's likelihood to have certain genetic conditions, ultrasound findings can conflict with genetic testing results, or when using parental carrier screening it may be difficult to determine the exact chances that a child inherits a condition from their parents. Tests may find a 'variant of uncertain significance' (VUS), a gene that may potentially be pathogenic

but could also be benign – hence its 'uncertain significance.' It is also very difficult to interpret whether these variants will present more severely or mildly in the child, and what that could mean for their medical needs or disability status (e.g., as discussed regarding sex chromosome aneuploidies). In general, even though scientists have sequenced the entire human genome, given the multitude of variations and mutations, it is not always clear how a gene or a particular permutation of genes may impact an individual. The clinical databases around how genes cooperate to influence a person's health and medical needs are being developed concurrently, alongside emerging new genetic technologies and the increased use of genetic testing in medical spaces such as fertility and reproduction, pediatrics, and cancer care.

As prenatal genetic testing becomes routinized – bringing with it the uncertainty and ambiguity that is inherent to genetic health data – there are more instances wherein potential parents can be faced with prenatal genetic insights that leave them in a difficult situation. Ambiguous prenatal genetic data can be challenging for several reasons; for example, parents may be unsure of how to prepare for raising their child or may be stuck deciding whether to continue their pregnancy if they are not in a position to raise a child with additional needs. As prenatal genetic testing quickly becomes a more routine part of reproductive care, these complexities and ambiguities in genetic testing results are increasingly present for patients (some of whom may be unaware that their pregnancies were genetically tested, due to insufficient patient education from non-genetics providers ordering NIPT).

Being able to help patient navigate these moments takes not only additional time from providers, but specialized skills and strategies. As physicians de-medicalize and shuffle the responsibility of ambiguous and complex patient cases to genetic counselors, we see how counselors' expert ability to absorb the influx of uncertainty is crucial to addressing patients' medical needs and reproductive values. Through shuffling and absorbing, counselors' role as indispensable healthcare providers, as well as their larger identity as a feminized profession, is made more meaningful in the context of routinized genetic testing that direly necessitates such expertise. Once again, as counselors absorb and re-medicalize navigating ambiguity, they elevate their expert roles and expand what it means to provide equitable patientcare in genomic medicine.

Doctors. While reproductive physicians accept uncertainty as a part of providing patients genetic testing, they did not typically engage with patients around interpreting ambiguity for reproductive decision-making. Rather, doctors focused on triangulating clinical data to better interpret uncertain results and illuminate possible medical routes forward. As when faced with tough socioemotional situations, doctors' primary concern remained 'objective' or 'rational' clinical interventions and medical next steps. They understood prenatal genetic results as a part of a wider constellation of medical markers about the pregnancy, working alongside counselors to 'piece together' findings from genetic tests, ultrasounds, and other pregnancy screenings to better understand a fetus' health state and plan pregnancy management. At times, doctors provided patients directive recommendations for clinical steps. Some also described providing patients with follow-up ultrasounds after receiving an uncertain genetic result as a way of symbolically indicating to patients that closer care is taking place, even if uncertainty may not necessarily be resolved. However, doctors overall admitted struggling when responding to uncertain genetic findings largely because ambiguous findings did not usually indicate apparent next steps for clinical

management – physicians' scope of medicalized expertise. Dr. Khoury explains how she approaches uncertain findings:

> "(Genetic data) very easy because it's quantitative and you have this mutation, and you can describe it in lay terms to anybody […]. But the issue becomes when that's the only objective evidence that we have. I know I can see this anomaly, I have this objective evidence, but how the phenotype or how the postnatal manifestation will be, that's very difficult, I cannot answer that. […] I have no problem talking about what's objective, and I will have no problem telling them that I don't know, we don't know, no one will know. […] Just having them understand how the gene is separate than the actual phenotype in some cases is what's needed. And that's why my genetic counselors are my friends; they have a lot of the support and the resources to spend time with families for that. […] So, I refer (patients) to communities, refer them to the literature that we have, and the objective evidence." (Dr. Khoury Interview 2021)

Doctors treated uncertain findings 'objective evidence' that needed to be understood and accepted accordingly. In cases of uncertainty, they were largely satisfied with communicating to patients that they 'did not know' what a variant might mean. While this may represent an honest approach to patient engagement, it often left patients lost in terms of how to understand their pregnancies, which significantly impacted the patient experience. In these moments, patients had to navigate the ambiguity in their prenatal genetic testing results via other providers and resources. Although doctors were well-prepared for clinical management when genetic tests presented clear evidence to act on, when data was uncertain, they de-medicalized its significance and readily referred patients to genetic counselors, shuffling the responsibility of these complex and ambiguous patient cases. Several physicians also expressed being less skilled at managing uncertainty in general. Consequently, doctors' routine workflow included sending 'complicated' cases to genetic counselors: "If they get a VUS […] that's going to go to genetics. To be perfectly honest with you, if it says, 'no variants,' then I'm good. The moment that it gets more complicated, […] like if an NIPT result is abnormal, the genetics referral will be automatic […]." (Dr. Das Interview 2021). In the large private hospital where ObGyn Dr. Das works, the shuffling of ambiguity related to genetic medicine is built into the healthcare system.

Given that doctors framed ambiguity an expected and objective part of genetics, expecting patients to accept that such data was necessarily uncertain or uninformative, they did not always recognize the larger medicalized implications this may have for patients. At times, there were negative consequences for patients when physicians' approached uncertainty as a routine clinical given rather than a finding that may impact patients' reproductive decision-making and broader experience. In comparison, genetic counselors (who receive specific training in counseling around ambiguous genetic data) were more cautious when presenting uncertain findings, expertly conveying and medically contextualizing the ambiguity without raising alarm about a finding that has no apparent clinical implications. Below, Dr. Scott, a perinatologist, describes delivering uncertain findings to a patient, not realizing the response the patient would have:

> "I had a patient whose first child was normal, her second severely disabled on a ventilator at home, and […] she wanted to do (a genetic) test to see if this third was affected. […] It just came back, whole exome sequencing, with one unknown variant. I thought she would

> be just thrilled that nothing popped up that was serious. But she was all concentrated on the variant of unknown significance. […] Anyway, she went back to her genetic counselor because I said, 'I don't have expertise in reading the sequencing test, you need to talk to your genetic counselor.' […] But obviously she got caught on that one sentence I put in her email that there was a bug variant of unknown significance." (Dr. Scott Interview 2021).

Where Dr. Scott included the VUS finding in her email to patients, many counselors would have perhaps delivered these results to patients differently, revealing why Dr. Scott ultimately turned to her genetic counselor colleagues for help. Here, it is also worth recognizing that as Dr. Scott shuffles the responsibility of specialized genomics and ambiguous genetic data to genetic counselors – that is, reading a whole genome sequencing result with a variant of uncertain significance – she admittedly notes that she does not have the adequate expertise to address the patients' needs around understanding these genetic results. Accordingly, as she shuffles these responsibilities to genetic counselors, she also uplifts their relative expertise in being able to engage patients and provide healthcare that she is not equipped to, illustrating the growing importance of the genetic counselor's expert role in delivering precision medicine.

It is not only that genetic test results can present ambiguity for patients, but there are also consequential decisions that need to be made by providers who order tests to mitigate the potential for uncertain results. This requires a thorough understanding of the patient's particular case, specialized genomics knowledge to interpret the patient's testing and technological needs, as well as familiarity with the various testing avenues that may be suitable. Physicians explicitly stated that they did not have this level of in-depth expertise around ordering prenatal genetic testing; it was too 'nuanced' and not pertinent enough to 'objective' medical work to comprise their scope of practice. As such, physicians turned to genetic counselors to manage uncertainty through the logistics of genetic testing according to various genetic technologies, laboratories, and clinical specificities. Doctors relied on genetic counselors to know which laboratories to order testing from based on a pregnancy's particularities, as ordering an inappropriate test or relying on the wrong laboratory for a specific patient case could lead to inconclusive findings and obfuscate patient care. Dr. Schuster, an MFM, explains further:

> "(Genetic counselors) have a more nuanced understanding of the capabilities of the labs […]. You can get an indeterminate NIPT where (the laboratory) can't read it. Often if you get it wrong between 10 and 12 weeks, there's less fetal DNA circulating, and in women who are obese there's a dilution effect so they can't get enough fetal fraction, and women who have lupus sometimes because they have some extra DNA circulating. So, my genetic counselors have relationships with each of those lab[oratories], and they can call the lab[oratory] and say this is indeterminate but is it just because it was not enough fetal fraction? Or were you worried about something, but you couldn't call it?" (Dr. Schuster Interview 2021).

While doctors handle uncertain findings by planning for prospective clinical management and treating ambiguity as medical evidence, when it comes to patient interactions and technical choices around testing, they depend on counselors to help select appropriate testing avenues, liaise with laboratories, and support potential parents in interpreting uncertainty and navigating complicated

decision-making. Being able to connect with laboratories to unravel the source of uncertainty or understand whether an inconclusive finding is indeed worrisome is central to subsequent medical care and reproductive decisions – and particularly valuable in a biomedical context where more patients are being prenatally tested, engendering more complex genetic data about pregnancies. However, even as physicians recognized counselors specialized expertise in navigating uncertainty, they de-medicalized the skills and technical logistics counselors needed to employ when helping patients understand ambiguous results and mitigate unknowns. Dr. Schuster, for example, goes onto explain that her counselors' labor on this front enables *her* to provide informed medical recommendations (e.g. further testing or pregnancy interventions) and appropriately plan for clinical care – though the counselor's expertise is not seen as medical itself but rather as 'relationship-building' with laboratories. Even though doctors willingly (and necessarily) shuffle complex and ambiguous cases to genetic counselors, and admittedly rely on counselors' expertise to carry out clinical interventions, they framed these responsibilities as tangential to medical work.

Genetic counselors' skills are often characterized as traditionally feminized labor, making the budding emphasis on their roles in precision medicine more meaningful. As seen Dr. Schuster's interview (as well as accounts presented from other doctors), genetic counselors' expertise around navigating ambiguity is often discussed as 'nuanced' work, and their skills are contextualized as 'relationship-building,' be it with patients or genomics laboratories and companies. These references to work that is 'detailed' or that represents 'soft' social skills typically characterizes feminized work, where the additional effort and specialized skills taken to carry out such expertise is not appropriately recognized (Hochschild 2011; Hochschild and Ehrenreich 2004; Salzinger 2004). However, that physicians not only shuffle these responsibilities to genetic counselors, but moreover see the necessity in doing so, (and that counselors strategically absorb these tasks, as discussed below), perhaps contributes to uplifting these traditionally feminized and under-valued skills in the field of reproductive genomic medicine. As counselors skills are better recognized through this dialectical shuffling and absorbing, their expert roles are also more centralized in addressing patients' needs, giving voice to a historically feminized profession that has been largely excluded from influencing genomic medicine. Accordingly, as genetic testing becomes routinized, it positions genetic counselors and their expertise as *invaluable* and *necessary* for equitable patientcare that increasingly involves managing uncertain genetic data, evolving technologies and technical considerations, and complex patient needs.

Genetic Counselors. With the influx of genetic data and its inherent complexity in reproductive medicine, counselors' highlight their expertise as increasingly critical to medical care. Counselors are especially adept at helping patients navigate 'grey' or ambiguous situations, with specific training that helps them build such expertise. They delve into uncertainty and provide clarity when possible, situating ambiguous findings in a patient's broader context and conducting in-depth research about the particular genetic variation as needed. However, uncertainty cannot always be resolved; in these instances, genetic counselors are stewards in helping patients navigate murky grounds and make reproductive decisions based on incomplete or indeterminate information. In this way, counselors emphasized the medical relevance of their expertise and underscored their ability to absorb complex and ambiguous patient cases as constitutive of equitable patientcare.

For counselors, who are trained to expect ambiguous genetic data and anticipate patients' related concerns, supporting patients through uncertainty often begins before testing has been ordered. As

such, part of genetic counselors' pre-test counseling focused on preparing patients for potential ambiguous findings, so they can decide whether to test based on the degree of uncertainty they want to contend with in their pregnancy. While doctors typically saw genetic tests as 'objectively' useful clinical data (even if they did return ambiguous results) and often recommended testing across patients, genetic counselors insisted that patients' consent to the extent of uncertainty genetic tests bring, offering patients more choice in the type of reproductive experience they were seeking to have. Counselor Emma describes a typical approach to managing uncertainty in a pre-test counseling context:

> "We do sometimes end up with uncertainty […]. We have a variety of strategies to try to help patients through that during pretest counseling. Just knowing before you do a test what your possible test results could be, psychologically, makes a huge difference. It helps you prepare for that idea. It gives you that active choice on whether you want to do that test or not. That is a big component of it." (Emma Interview 2021)

Emma emphasizes that VUS' findings in prenatal genetic testing are often benign and unnecessarily worry pregnant patients. Counselors' attention to whether and how patients' receive uncertain prenatal genetic data also adheres to patient-centered socioemotional needs and represents a key part of genetic counselors' clinical engagement approach (in contrast to many physicians, who expected patients to accept the 'objectively' unknowable qualities of some genetic variations). As such, once test results were available, genetic counselors described being very cautious about presenting uncertain findings to patients because of how this information may add stress to expecting parents' experiences and impact their reproductive decisions. Many counselors described their approach as "innocent until proven guilty" – they would not discuss a VUS as concerning unless it was scientifically evidenced, in extant research and the clinical database shared across genetics practitioners, to result in health changes. They were primarily concerned about patients' emotional responses to ambiguous data: "We don't want them to jump to decisions because most VUS end up being benign. But it's really, really, really hard to convince a couple that a finding doesn't mean anything, especially if somebody already told them, 'Hey we found something.' That's really hard to undo (Tanya Interview 2021)." Genetic counselors paid close attention to how ambiguous information was delivered because of the substantial impact it can have on patients' pregnancy decisions. As Tanya notes, helping patients navigate uncertainty can also represent 'damage control' when other providers have reported ambiguous or indeterminate findings to patients without consideration for how this influences their pregnancy experience.

Genetic counselors absorb uncertainty navigation from physicians as a part of providing patients specialized genetics care, which is a defined part of their trained expertise, and is increasingly pertinent for patients' medical needs as genetic testing becomes a routine part of reproductive care. Along these lines, counselors had various specific strategies to mitigate uncertainty for patients as they pursued prenatal genetic testing. For instance, counselors were trained to interpret uncertain results within a patient's personal and medical context, and in doing so were attentive to their larger pregnancy experience and potential harmful consequences that may arise from being faced with ambiguous prenatal genetic test results. Recognizing that patients may subsequently make consequential pregnancy-related decisions based on ambiguous genetic data, several genetic counselors illuminated uncertain findings only when potential clinical actions existed. They would also translate a VUS to speak to a patient's particular circumstances and reproductive priorities

(e.g., continue versus termination preferences, other concerning fetal medical markers). In doing so, counselors re-medicalized navigating uncertainty as integral to patients' clinical experiences and reproductive values; they situated absorbing this expertise in providing equitable and responsive care to pregnant patients faced with increasing amounts of genetic data. Skylar, who works with many patients with lower health literacy at a large community hospital, illustrates how she situates uncertain findings in a patient's particular context:

> "The most recent (case) I had was such a nightmare. A young patient, she was a minor, and she hadn't been given the privilege of a good education, she was still in high school. And we had a VUS come up […]. The phenotype of the fetus had some mild stuff, […] I'm surprised the lab reported it in a prenatal setting[59] […]. So, I would tell her I'm 95% sure that this (variant) does nothing. […] That's the logic I approach it with, I look at it as much as possible and try to figure it out, what are we going to do with this? And the answer is nothing. Do we want a patient considering termination based on ambiguous finding? Probably not. Is this a desired baby otherwise? Are they comfortable with what's on ultrasound otherwise? So, if this is not an actionable finding […] I am extremely reassuring as long as I feel like it's medically appropriate. […] I try to keep patients optimistic and empowered and continuing to bond with their pregnancies if they are deciding to continue […]. It's my responsibility to help them cope with this information as much as possible […]." (Skylar Interview 2021).

With patients' experiences at the top of their minds, genetic counselors are deliberate in how they present uncertain results as medical information and help patients navigate this territory. They see navigating ambiguous genetic data and complex patient engagement as a fundamental aspect of their expert role. In particular, counselors seek to mitigate ambiguity and related anxieties (rather than leaving patients to understand this as a 'given' in genetic data), recognizing that uncertain genetic data, even if it has no known health or medical implications, can influence major decisions patients make about their pregnancy. This is especially important when a pregnancy is wanted or when expecting parents have expressed the desire to continue their pregnancy. As Skylar notes, her approach is to understand the uncertain variant as best possible and present information to a patient based on whether the variant suggests potential future clinical actions. In this way, genetic counselors interpret ambiguous prenatal findings in the broader context of each patient, considering whether they want to continue their pregnancy and whether other markers about the fetus indicate healthiness in the way that the variant is discussed. They re-medicalize this approach as essential to equitable patientcare, as it fundamentally centers patients' needs and reproductive priorities.

Not only do counselors appropriately situate uncertain genetic data in patients' personal and medical contexts, but they also dedicated substantial effort to background research on genetic variants and liaised with specialists and laboratories to connect and cooperatively interpret any relevant information about unclear genetic findings. Accordingly, uncertainty was not taken for granted as an objective medical fact; genetic counselors used their expertise to medically transform

[59] In the prenatal genetic context, many laboratories do not report VUS' in patients test results, unless the VUS has been well-established in the extant literature as having some degree of health or medical significance. This practice among laboratories is to further protect patients and providers from 'information overload,' as most VUS' in the prenatal context are benign and inconsequential for an individual's health and medical needs.

and interpret ambiguity meaningfully, to deliver patients findings that are holistic, well-informed, and interpreted in terms of relevance for their specific pregnancy. Along these lines, counselor Mara explains that her first response to receiving a VUS finding is turning to additional research: "The first thing I do is talk to our geneticists […] and try to get an idea of their thoughts about it. Then I try to get as much literature (on the variant). And then I'll call the lab[oratory]. So, I try to do my homework first before calling the patient […]. I want to be prepared ahead of time when communicating uncertainty (Mara Interview 2021)." As seen in Mara's account, to make uncertain genetic data more significant for a particular patient, counselors absorb the additional labor related to research and relationship-building with other experts. Even as genetic counselors take a nuanced and emotionally responsive approach to managing uncertainty, they prioritize presenting scientifically sound and accurate information to patients (as Skylar also notes above, she is only reassuring when medically appropriate), further illustrating the medical significance of their specialized genomics expertise.

When patients were grappling with reproductive decisions in light of uncertain prenatal genetic findings, genetic counselors would employ strategies to present various possible outcomes and ways forward to patients, in order to help patients gauge their own comfort-level and preferences around the ambiguous information about their pregnancy. Specifically, if they knew that a patient's prenatal genetic test had returned uncertain findings, many genetic counselors would discuss potential clinical management routes with specialists ahead of meeting with the patient (e.g., conducting a follow-up fetal echocardiogram). They would present this to patients as a way to 'act on' uncertain results, or an option for a path forward, guiding decision-making without being directive. I also observed many genetic counselors interpreting patients' reproductive preferences based on cues in their consultation, which they would then use to present various hypothetical scenarios that a patient could respond to (a strategy counselors also referred to in their interviews; Genetic Counseling Ethnography 2021). Below, counselor Whitney describes relying on cues to ascertain patients' pregnancy preferences, while also presenting them with decisions and paths that other patients in their situation have taken. Counselors described this strategy as clarifying tangible ways forward for patients struggling to understand the uncertainty in their pregnancy, and in doing so helping patients decide where their own preferences land:

> "We can help with decision making based on what they express their desires and wants and needs to be […]. Like if they say, 'I don't need to find out about every single genetic condition in my pregnancy' with certainty, then we can say, 'it's reasonable to decline the amniocentesis' […]. While I wouldn't ever say, 'I recommend you get an amniocentesis' I can walk people through examples of what others do in a similar situation. Patients can compare those parallels to see if that's what they want to do too." (Whitney Interview 2021).

Genetic counselors expertise in navigating uncertainty is important to patients' medical experience in myriad ways. Counselors skills around relationship-building are not only important to liaising with other specialists, but they also maintained close ties with former patient families, patient resource centers, and patient support groups. They regularly referred patients to others who faced similar uncertainty or to relevant disability communities. This not only gave patients an understanding of their options but was critical to education around disability and diverse existence, as it enabled expecting parents to get a sense of their child's potential life experience and connect

with other families in their position. As such, in taking on the navigation of uncertainty and re-medicalizing its significance within routinized genetics, counselors also brought more equitable perspectives on various existences and empowered patients' range of reproductive choices.

Overall, genetic counselors carry out crucial relational work as they translate uncertainty for both doctors and patients, helping to make ambiguous genetic data clinically actionable and approachable while also situating findings (and implications, if any) within patients' personal and medical contexts. They also collaborate alongside other experts, bridging scientific research, specialist perspectives, and laboratory analyses in the way they make genetic results meaningful and counsel patients. This ability to connect various sources of information for a more robust understanding of how genetic data can inform prenatal clinical care represents an integral expertise. In particular, in a biomedical setting where genetic data and its inherent complexities are steadily more present, counselors absorbing the navigation of ambiguity from doctors, and encompassing it firmly within their defined scope of expertise, helps reposition them as a valuable expert role supporting both patients and providers. As they re-medicalize navigating ambiguous and complex prenatal cases as key to patient empowerment and clinical care, counselors importantly redefine delivering equitable genomic medicine to include this tenet.

The way that genetic counselors navigate uncertainty and complexity also requires substantial emotional labor to build rapport with patients, maintain connections with patient communities and families, and develop relationships with other specialists and resource providers. Further, not only does additional in-depth research involves more labor on the counselors' part but this work takes particular attention to detail to interpret ambiguous findings and situate these in each patient's personalized context. As discussed, such skills have traditionally been characterized as feminized work, unpaid and undervalued as such. However, in the precision medicine era, this 'feminized' expertise has emerged as critical to adequately addressing patients' needs in a way that is both socioemotionally responsive and attentive to heightened reliance on genetic technologies that render some uncertain data. Taken together, we thus see how this dialectical shuffling/absorbing unfolding in the current sociomedical context not only uplifts the counselor's expert role but also revalorizes skills that have long been considered 'feminized' and underrecognized in medicine.

Final Thoughts on Transforming Experts and Expertise in Reproductive Genetic Medicine

Expert roles and relationships to one another are far from stagnant. How experts define their scope of work and specialized expertise, and the resulting dynamics they share with other professionals in their field, shift based on the demands of a particular moment or context. These transformations can often be opportunities for reshaping a field – its values and societal purpose, the types of expertise it considers meaningful, and the experts positioned at its forefront (Abbott 1983; Eyal 2013; Navon and Eyal 2016). This chapter has illustrated how these processes play out in reproductive genomic medicine, specifically between reproductive physicians and genetic counselors.

Doctors shuffle specialized genomics responsibilities and related in-depth patient engagement to genetic counselors. Doctors de-medicalize these responsibilities as outside their scope of medical practice, and in some cases as uninteresting to their professional occupation. As they shuffle, doctors also frame genetic counselors as better suited to take on this type of specialized and in-depth work related to reproductive genetic testing. Specifically, two key aspects of specialized

genomics that doctors shuffle to genetic counselors are patient care responsibilities requiring socioemotionally responsive counseling and the navigation of uncertainty or complexity in genetic testing processes. For their part, genetic counselors strategically absorb these responsibilities, and encompass these within their defined scope of expertise. And, given the sustained patient-centered care movement and growing healthcare focus on genomic technologies, counselors' work becomes more clinically relevant and central to emerging patient needs. As they absorb, counselors strategically position themselves as *the* experts needed to take on these responsibilities, which they re-medicalize as integral to equitable patientcare and reproductive decision making. Indeed, we see that prenatal genetic medicine cannot happen as it does without counselors, which perhaps expands the understanding around what (and whose) expertise constitutes supporting medical care in genomics.

This shuffling and absorbing process among doctors and counselors is dialectical, in that it requires both groups to participate in their respective part of the practice. However, it is not mutually or intentionally cooperative, as much of the shift in responsibilities is based on each group's self-interests and self-defined scope of practice, rather than an expressed desire to effectively cooperate with other professionals in their field. Still, shuffling and absorbing is transformative in reproductive genomic medicine, where genetic counselors, a historically less powerful group compared to physicians, are being elevated as critical healthcare providers, demonstrating their willingness and ability to carry out expertise that are especially salient to prenatal patients' medical needs. Moreover, counselors' increasingly relevant and necessary expertise represent skills that have historically been considered feminized; elevating such expertise and foregrounding these experts in genomic medicine, where voices of feminized groups have long been excluded and undervalued, could meaningfully transform the values around patientcare in this field.[60]

While this chapter has focused on illustrating how genetic counselors expert roles and expertise are being revalorized given the current sociomedical context, I do not intend to convey that doctors are not essential to delivering high-quality care for prenatal patients pursuing genetic testing. Of course, doctors are critical to addressing patients' needs in reproductive genetic medicine; it would be naïve to argue otherwise. Rather, this chapter shows how expert providers need to (and often do) work in collaboration with one another to meet shared objectives (e.g., around patientcare), and should accordingly be recognized for the expertise and roles they occupy. In this case, the dialectical shuffling and absorbing process illuminates how such cooperation takes place (albeit largely unintentionally), and how patients indeed benefit from this changing workflow among doctors and genetic counselors, with each expert better able to focus on their specific expertise and scope of patient engagement. However, it remains that genetic counselors are still not adequately recognized for their contributions to patientcare in prenatal genetics. Counselors took on substantial labor that was not being formally acknowledged. Recall, even though genetic counselors cannot order and bill for genetic tests per insurance policies (a core issue the NSGC Act is trying to rectify), many physicians relied on them to order the correct tests for prenatal

[60] Seim develops the concept of burden shuffling, with a robust discussion around how this takes place in emergency medicine and paramedical services. This chapter builds on Seim's concept of burden shuffling, adding how professionals can strategically absorb 'burdensome' work being shuffled to them. The pairing of shuffling alongside absorbing is significant, as it illustrates how such dialectical processes can contribute to important transformations in expert roles and fields, in some cases (including that of genetic counselors and physicians in reproductive genetic medicine) changing authority structures and elevating less powerful professional groups. (Seim 2017, 2020)

patients, with counselors doing so yet billing under physicians' names (and therefore not being adequately compensated). In several instances, physicians did not check these orders before they were placed because they admittedly had less understanding of which genetic tests were needed. Physicians' less thorough understanding of genetic testing was also reflected in counselors having to 'damage control' or walk back misinformed counseling physicians provided to patients, describing these moments as especially challenging and deleterious to the patient experience. In these instances, we see how counselors are performing specialized genomics expertise that is not being legitimized within their professional landscape. As it would be for any profession, it is critical to genetic counselors that they receive recognition and compensation commensurate to their expertise and contributions.

The Act presents a critical opportunity for genetic counselors and broader equity in the field of genomic medicine. If passed into law, this NSGC Act would allow genetic counselors the recognition and financial compensation based on the many responsibilities (e.g., ordering genetic tests, engaging in-depth patientcare) that they already take on. This Act could transform insurance policies and healthcare to allow greater access to genetic counselors as a critical group of experts, which is essential in today's context where there is a growing reliance on genetic testing yet a persistent need for genetics professionals who can appropriately use and interpret these tests. The development and routine implementation of medical genetic technologies continues to outpace the experts ready to counsel patients and providers on how to use these tools. Per their 2019 workforce report, The National Society of Genetic Counselors estimates that there is 1 genetics professional per 300,000 patients, representing a dire shortage of genetic counselors in the United States (National Society of Genetic Counselors 2019). The NSGC also projects that the demand for genetic counselors will continue to grow by 28% from 2020 to 2028, based on how genetic testing technologies are being rapidly routinized across medical specialties (Healthcare Management 2023).[61] Accordingly, in their Act, genetic counselors argue that better empowering them as professionals meets the needs of patients, in terms of facilitating access to providers specialized in genetic testing. As doctors and counselors shuffle and strategically absorb specialized genomics responsibilities, and as counselors further mobilize around their Act, we could see new possibilities for how genetic counselors are valued in healthcare. The Act may enable more diverse perspectives (i.e., from counselors) to influence how genomic medicine and related patient care are carried out 'equitably,' uplifting expertise specifically suited to patient needs in precision medicine era. (Higgins 2021; National Society of Genetic Counselors 2021b, 2022)

The NSGC Act's potential for illuminating new and improved modes of equitable care, which foremost centralizes patient needs, is particularly significant. Given the rise in new medical genetic technologies, uplifting expert roles and expertise suited for the moment can have notable impacts on disability justice and inclusion, reproductive empowerment, health identities and diagnostic categories, and social and individual well-being. It is imperative to recognize that the roots of genomic medicine can be traced to eugenics and ableist constructions of disability that obscure social causes of health disparities (Phillips 2020). Uncritical modes of implementing genetic

[61] Similar to genetic counselors, The Bureau of Labor Statistics estimates that the employment of geneticists (physicians trained in the genetics specialty) will also grow by 27% from 2020 to 2030, a rate that is much faster than the average for all occupations (Bureau of Labor Statistics 2023). Together, these statistics illustrate the heightening demand for genetics experts who are trained to appropriately implement genetic testing and thoroughly inform patients of the process and possible results.

science and technologies can perpetuate ableist ideals, privilege technological access (and related social and health benefits) for those who are well-resourced, essentialize genetic conditions, and stigmatize or target communities based on genetic findings (Bridges 2022; Navon 2019; Roberts 2012; Timmermans and Kaufman 2020). Along these lines, 'genetic determinism' – the idea that health and social outcomes can be essentialized to one's genes – remains a main issue regarding how health is conceived in precision medicine. There are also concerns about routinized genetic testing engendering an overly individualized approach to medicine, placing unreasonable accountability on patients to navigate healthcare based on genetic markers while overlooking critical gene-environment contexts (Rose 2001, 2008; Shostak 2003; Timmermans and Kaufman 2020:593). This is especially pertinent to prenatal genetic testing, where pregnant people can be socially pressured into genetically testing or even terminating fetuses based on expectations to manage their families' health (Meredith et al. 2023; Rapp 1994, 1998; Rayna Rapp 2001).[62]

As more conditions (e.g., cancer, alcoholism, schizophrenia) become reconstructed as genetic diseases, the need for experts who are trained to carry out patient engagement and responsibilities around genetic testing through equitable approaches becomes ever more pressing (Lippman 1992). Providing patients sufficient access to genetic experts who can provide critical and informed care is of utmost importance to acknowledging and proactively addressing these inequities that lie at the foundation of genomic medicine. As the NSGC insists in their Act, genetic counselors are specifically suited to filling this role, as their training and education involves understanding the problematic roots of genomic medicine and the ways their counseling strategies can be adapted to undoing or mitigating some of these harms. While access to genetic counselors is not an exhaustive solution to inequity in genetic medicine, it is a key part of building this discourse and practice. (Higgins 2021; National Society of Genetic Counselors 2021b, 2022; Master's Programs Ethnography 2021).

As the NSGC Act gains more traction, with support from a variety of stakeholders (increasingly including physician communities), many genetic counselors are optimistic for the possible changes it can bring. However, as we wait to see how the Act unfolds, it is worth keeping a critical eye toward developments in new genetic technologies and the myriad ways our social arrangements, especially with regard to equitable and informed healthcare, are structured around these changes. With prenatal genetic testing being more commonplace than it has ever been (largely enabled by NIPT), and patients gaining more access to these tools, it is especially important to underscore how interactions around these technologies bear implications for how expertise is developed, which expert roles are considered meaningful to technological innovation, and resulting experiences for patients. As they are embedded in particular sociomedical contexts, these dynamics influence what types of knowledge and expertise are valued in the process.

That expert roles can transform given contemporary sociomedical contexts entails that knowledge-making (based on whose expertise is valued at a time) is consistently changing as well (Eyal 2013; Navon and Eyal 2016). Thus, there are ongoing opportunities for change when it comes to experts and expertise, as well as the possibility to observe how these changes impact a particular field and

[62] In prenatal testing, these pressures are compounded by market forces that directly target anxious patients with genetic testing products, as was seen with the early marketing around non-invasive prenatal testing that pitched the product to expecting parents as a 'gender test' or 'the test for Down Syndrome' (Minear et al. 2015).

those it serves. It is important to study these changes which highlight moments of resistance and reclamation in science, medicine, and technology; for example, in the case of doctors and genetic counselors we may be witnessing a historical moment wherein a less powerful and traditionally feminized expert group could be transforming and gaining more influence over a field that has long excluded their voices. When it comes to rapidly emerging prenatal genetic innovations, shifts in expert dynamics impact the broader social experience around how these technologies are developed and practiced. Understanding how these processes among experts unfold is essential to foreseeing and addressing the consequences for individuals' reproductive medical experiences and social equity, as decisions around health and existence are increasingly intertwined with genetic technologies to determine whether and how various individuals come into being.

CHAPTER FIVE

Conclusion

Precision medicine has promised 'better' health through attention to risk on an individual level. Using personalized markers, such as knowledge about their particular genetic mutations, individuals can now access treatments especially suited for their health needs, manage their risks before they exacerbate into symptoms, and access preventative care for diseases that have yet to manifest. The goal: to enable longer lives, develop more effective treatments, and reduce healthcare costs by preventing or identifying and treating disease earlier.[63] Key to standardizing precision medicine has been genomic knowledge. Armed with findings from the Human Genome Project (completed in 2003), scientists pinpointed the genes found in human DNA – discerning the typical genome from 'abnormal' variations – and marshalled these insights to create more precise and sensitive genetic technologies. These precision medicine tools have been especially transformative in the medical management of reproduction. Hopeful parents can test for conditions they may be carriers for, adjusting their reproductive plans to avoid passing on particular genes or having better insight into their child's possible health needs. Individuals using in vitro fertilization might genetically test and select 'the best' embryos for implantation. Or, as this dissertation has explored, parents and providers frequently pursue prenatal genetic testing to understand fetal genetic health and attend to pregnancies accordingly. (Lippman 1992; Mukherjee et al. 2022; Navon 2019; Obama 2015; Rose 2001, 2008; Timmermans and Kaufman 2020; White House 2015b)

In many ways, this dissertation picks up where Daniel Navon left off in *Mobilizing Mutations*, where he forecasts the implications of genetic testing in prenatal medicine and tells us that "genetic mutations can reshape what it means to be ill, different, and ultimately, human" (Navon 2019:314). Indeed, these shifting conceptions have been the focus of my exploration. While prenatal genetic testing certainly enables some in-utero interventions and targeted pre- and post-natal attention, it often prompts decisions around termination or continuation based on a fetus' possible genetic conditions. As such, compared to other medical fields, using genetic tools to manage reproduction raises critical and unique questions because it fundamentally bears on *who is brought to life* based on their genetic constitution. Reproductive genetic tools transform how we societally think about disability, difference, and what makes for a meaningful life. With prenatal genetic technologies prying open the door to more genetic variations being identified and medicalized in-utero, our collective calculus around what represents a 'normal' or desirable life is continually shifting. And with tools like NIPT becoming all but routinized, more expecting parents are learning of fetal genetic differences earlier in pregnancies, identifying a growing list of variations (some without

[63] Precision medicine has also had a significant impact on oncology, for example. Increasingly, individuals are getting genetically tested to understand their risks for developing particular cancers or diagnose a cause for their cancer. Those with cancer-causing genetic mutations can access risk-reducing prophylactic treatments (e.g., women with BRCA1 or BRCA2 mutations often surgically remove breast tissue, ovarian tissue, and fallopian tubes to reduce their risk of cancers in these areas), direct treatments based on the genetic nature of their cancer (e.g., cancer patients with certain genetic mutations can access PARP inhibitor medications to reduce cancer recurrence), and engage in ongoing targeted surveillance to monitor their health so that potential disease can be caught and treated as early as possible. These medical advancements, largely enabled by genomic knowledge and tools, have had life-saving effects for many cancer patients and those at risk for developing cancer. Genomics has been critical to care in other medical fields as well, including pediatrics, reproduction, multi-faceted developmental disabilities, and hematology (e.g., sickle cell anemia).

notable medical implications, like SCAs), and contending with what this information actually means for their future child's health and disability status. The concepts of health and risk, disability, and socially equitable healthcare hang in a delicate balance alongside quickly developing reproductive genetic innovations. (van der Meij et al. 2022; Meredith et al. 2023; Minear et al. 2015; Navon 2019; Rapp 1994, 1998)

The first empirical chapter traced how innovations in prenatal genetic technologies have transformed societal notions of risk tolerance, health responsibility, and disability over the past six decades. It asked who is brought to life as prenatal genetic tools become sharper and more routinely available, illustrating how these testing practices shape our conceptions around disability and meaningful existence. As courts adjudicated Wrongful Birth and Life cases, they continually underscored the imperative to use these tools to 'judiciously' reproduce genetic normalcy and able-bodied biocitizens. With more moralized expectations on parents and providers to employ these tests, courts gradually shaped a sociomedical culture that is less tolerant of technological error and genetic risk. Interpreting the 'appropriate' use of prenatal genetic technologies, courts not only established societal parameters around how this testing should be used to preclude births of those with disabilities and genetic conditions but also grounded the private responsibility to manage health toward this end. As a consequence, disability was transformed into an unwanted existence and a legal injury, with private compensation for affected families standing in the place of meaningful social supports. While the legal system is one of many essential social institutions, its judgements notably shape medicalized interactions around new technologies (e.g., outlining malpractice) and thus establish a precedent for broader sociocultural attitudes regarding how genetic tools *should* be used to prevent lives considered 'wrongful' and regretted.

The second empirical chapter broached further into how notions of disability and unwanted existences are socially construed and sharpened, as genetic tools reveal more variations among individuals. Focusing on sex chromosome aneuploidies, it questioned how diagnostic categories are being reconstructed and expanded in prenatal genetic medicine, showing how even minor genetic differences, at times with no health implications, are cast as pathological medical conditions. Here, the implications of NIPT as 'the gender test' were visible, as the routine (and often uninformed) use of this tool to reveal fetal sex chromosomes spiraled into medicalized efforts to align those of so-called aberrant sex with stereotypical expectations of cis-gendered identity. Reproductive physicians and genomic researchers pathologized gender to delineate SCAs as an 'abnormal' existence and create urgency around treating and funding research about these variations. Despite the plethora of research establishing variability in sex and gender, physicians in clinics and research settings framed individuals with chromosomes outside XX or XY as abnormal – requiring clinical interventions to conform their gendered development with a medicalized determination of their genetic and embodied sex. In their efforts, physicians also medicalized traits and characteristics related to SCAs – such as height and infertility – much earlier in one's life course, framing these issues as medical concerns for fetuses in-utero where they otherwise may have never significantly impacted one's life. As a result, reproductive decision-making becomes far more complex and complicated, as parents struggled to interpret the medically relevant consequences of SCAs. As Navon notes, "finding abnormal genomes is becoming relatively easy. Helping people understand and make decisions based on prenatal genetic testing results with uncertain implications, by contrast, represents a largely overlooked challenge" (Navon 2019:302–3). Overall, we see how 'objective' medical science (e.g., diagnostic categories) is in

fact socially contoured. When perpetuated by experts, these social biases (e.g., regarding gender) can be mobilized to construct diagnostic categories that increasingly pathologize genetic differences and narrow the parameters around a normal or desirable existence.

The final empirical chapter delved deeper into the role of experts interpreting prenatal genetic tools and the 'best practices' for their implementation. It illustrated how reproductive physicians and genetic counselors dialectically coordinate and reorganize their work around prenatal technologies, transforming what it means to practice genomic medicine in the process. Doctors shuffled specialized genomics work they considered 'non-medical' to counselors, who strategically absorbed these responsibilities as a core part of their expertise. In absorbing specialized genomics responsibilities –such as socioemotionally responsive care and navigating uncertainty– counselors re-medicalized this work as not only necessary but critical to delivering equitable reproductive healthcare. In a context where we increasingly rely on genetic technologies and expect patient-centered care, counselors' expertise and approach to providing care alongside physicians was particularly salient to addressing patients' emergent needs. As a result of the shuffling and absorbing among experts, we see a redefinition of what it means to practice accessible and equitable genomic medicine, with counselors gaining more centrality as a valuable provider. Moreover, as a historically feminized and less powerful expert group, counselors (especially as they cement their efforts in the NSGC Act) could be influencing the values of a medical field that has traditionally excluded them, perhaps shaping how genomic medicine can integrate more socially equitable terms moving forward.

Reproductive genetic innovations profoundly impact how we understand human existence and disability. The increased reliance on these tools emphasizes responsibility toward managing health as a societal obligation. And even though genetic technologies are subjectively interpreted and acted upon, these insights ultimately aid the production of knowledge that comes to be taken-for-granted as medical fact. Underscoring the ramifications of how we use new genetic technologies, I outline several tangible recommendations to improve expert capacities and revise clinical practices that could enable a more equitable approach to prenatal genetic testing.

First, there is a need for more clinical genetics expertise and reproductive justice-informed genetics training for providers who consent patients to prenatal genetic testing. Relatedly, medical insurance policies should be revised to enable better access to balanced, comprehensive clinical genetics care. A readily actionable way to accomplish better access to genetics expertise and comprehensive patient education is enacting the NSGC's HR2144 Act, which would position counselors as providers under Medicare and Medicare and broaden patients' ability to benefit from their expertise (National Society of Genetic Counselors 2019, 2021b). This increased professional autonomy for counselors could also encourage more individuals to pursue this career, helping address the current critical shortage in genetic counseling experts. Relatedly, all medical professionals need to build capacity around critically implementing testing technologies with disability and reproductive justice foregrounded. There needs to be greater focus on patient consent through ensuring workflows between physicians, nurses, and counselors prioritize thorough pre-test genetic counseling as the norm. Providers, particularly doctors, need better training (and accountability) around giving balanced and comprehensive information about disabilities, equipping potential parents with supportive resources in equal part to informing them about the

medical challenges that may accompany a particular condition. (Markens 2013; Meredith et al. 2023; Minear et al. 2015)

Currently, commercial genomics companies– with their hefty investments, financial resources, and political sway– substantially define how NIPT is being incorporated into public health infrastructure (Coalition for Access to Prenatal Screening 2023; National Council on Disability 2019). Accordingly, second, we need patient communities to lead how prenatal tools like NIPT are equitably integrated into public healthcare. While expanding access (as genomics companies argue) to these technologies is important for patient equality and innovation, there can be harmful pitfalls to uncritically routinizing testing (van der Meij et al. 2022). As such, on a state-level, there needs to be more representation from patient advocates and disability communities when building prenatal genetic tools into public infrastructure, ensuring these technologies are used in alignment with social inclusion and tolerance for diverse existences. States should intentionally include and center these voices in their design of prenatal screening programs.

Third, there needs to be more discernment from experts, and optionality for patients, around which genetic variations we pathologize as medically relevant conditions. While technologies like NIPT will be able to identify more genetic differences with ease, it is up to those who develop and implement these tools (e.g., genomics researchers, innovators, and medical practitioners) to develop parameters around which variations comprise medical concerns that need to be tested for and presented to patients as *health* conditions. Further, clinics' and laboratories' testing protocols should build in more optionality around which variations, or categories of diagnoses, patients would like to test for (if any). Based on their reproductive priorities, patients may find it worthwhile to distinguish, for example, between conditions that lead to physical versus intellectual disabilities or those considered life-limiting versus life-threatening. More research into the frameworks people employ to define the varying ways a disability or genetic difference affects one's life would be useful in defining protocols for such optionality in prenatal genetic testing.

Still, there is no replacement for building equitable social structures and inclusive culture alongside developing genetic technologies and refining their implementation. States need to account for those with varying needs, consistently optimizing infrastructure, and providing resources so that individuals can participate meaningfully in socioeconomic life despite embodied challenges they may face. This also mitigates pressures on individuals (particularly those who are pregnant) to use genetic tools to preclude certain births and pushes societies closer to a justice-based approach to reproductive choice. Inclusive culture and accessible structures not only benefit disabled communities but enable society at-large to flourish with greater collective potential and participation.

Finally, with the accelerating pace of genomics innovations, especially in reproductive technologies, there are several areas necessitating further research. Notably, there is a need for more research documenting the systematic ways that anti-disability rhetoric is intertwined with medicalized implementation of reproductive genetic technologies. Meredith et al.'s 2023 study marks one of the more robust and up-to-date analyses of reproductive physicians' biases when consulting with patients who receive prenatal genetic testing findings (Meredith et al. 2023). It highlights how anti-disability prejudice is institutionalized in medical practices, as doctors present genetic conditions and disability (e.g., Down Syndrome) as tragic and unwanted, neglecting to

provide patients with supportive disability resources while stigmatizing the reproduction of families with such conditions. More research should build on this work to illustrate the prevalence of anti-disability biases across reproductive practices and providers. Here, it would also be worthwhile to understand how prenatal genetic findings, especially providers' approach to presenting results, impact parents' reproductive decision-making. While there has been work broaching this issue in the past (Asch 1999; Natoli et al. 2012; Pivetti and Melotti 2013; Rapp 1994, 1998), there are fewer contemporary studies systematically documenting the relationship between prenatal testing practices and termination patterns. Such work could illuminate sources of societal or institutional pressure on individual reproductive decisions and allow a more thorough understanding of disability acceptance. It is imperative that this vein of research is pursued within a reproductive-justice framework, being careful not to undermine parents' reproductive choices around termination or continuation of pregnancies (whether or not they are affected by genetic conditions and disabilities). Together, this research agenda can suggest evidence-informed recommendations that improve clinical protocols and disability education.

Scholars can also examine the intersection between new genetic technologies and commercial strategies in more depth. How does the commercial development and marketing of these tools shift broader culture and public attitudes around using testing as a moral or societal obligation? What are current consumer views around prenatal genetic testing and how this relates to privatized responsibility for family health management? How do commercial strategies to provide widespread access to testing shape perspectives (both on the State and individual levels) around disability and public supports for those with diverse needs? NIPT in particular highlighted the coalescence of politics, commercial interests, and biotechnology, as genomics companies who developed the tool not only marketed it intensely to patient-consumers and providers but formed a lobbying movement through CAPS to direct more public funding toward routinizing prenatal testing (Coalition for Access to Prenatal Screening 2023; Navon 2019:300). However, NIPT is not the only 'big business' in reproductive genetics; research on the commercialized contours of genetic testing might also consider direct-to-consumer genetic carrier testing, polygenic embryo screening (which is marketed directly to consumers, but must be ordered by providers, like NIPT), or boutique for-profit practices allowing for 'designer babies' and private use of genome editing. In a privatized health landscape, each of these technologies can reveal the effects that commercial practices have on disability stigma, technological imperatives, and responsibility for health and risk management. It is important to understand where these commercial efforts around innovations (and their power to influence sociopolitical and financial tides in healthcare) are bringing us, and purposefully ensure there is a meaningful role for health advocates and patient communities in shaping this direction. (Mukherjee et al. 2022; Roberts 2012)

Lastly, while my analysis of reproductive genetics tools was grounded in the history around eugenics, it is worth exploring whether eugenics ghosts are as prevalent in other applications of precision medicine. For example, do concerns around eugenics, genetic essentialism, and disability marginalization emerge in fields like oncology, hematology, or pediatrics where genetic tools are being similarly routinized? If not eugenics, what are the historical 'ghosts' or contexts informing the use of precision medicine tools in these spaces? As scholars have already suggested, ethnoracial determinism, biases against those of color and from low-income backgrounds, and unethical clinical research practices are all issues that may emerge in these inquiries (Phillips 2020; Roberts 1993, 2012; Timmermans and Kaufman 2020). These studies would be useful to a broader

understanding of social equity issues in contemporary medical genetic testing and could bolster targeted revisions to practices within each medical field.

Reproductive genetic technologies will not disappear from our sociomedical practices. They are indeed beneficial for many providers and patients, who can use genetic information to make decisions aligned with reproductive priorities, family visions, and optimal clinical care. Given this demand and utility, these technologies are being developed at a faster pace today. In 2019, Illumina Inc. released the next iteration of NIPT – VeriSeq NIPT Solution v2 – that can test for more fetal chromosomal conditions with heightened specificity and sensitivity. The test goes beyond its predecessor in identifying trisomy 21, 18, 13, and some SCAs, to now look for rare autosomal aneuploidies, more SCAs, and partial duplications and deletions of genes (Illumina 2023). With these innovations comes an ever-increasing reliance on genetic insights to make reproductive and medical decisions – a more complex endeavor as more genetic differences are brought to light without a full clinical understanding of how to interpret and act on these results. Given the shadow of eugenics in precision medicine (Phillips 2020), it is critical to understand the objectives behind these rapidly emerging technologies, unraveling which of these tools are being routinized in healthcare and at what cost. In reproduction, research must inform practices that harness these tools' technological benefits without amplifying their technological imperative. This can make all the difference in ensuring we do not materialize futures that are harmful to those with disabilities and other marginalized identities, singularly dictated by those with more professional and political power, and intolerant of one's range of reproductive choices. Most of all, continuing to push for more equitable technological futures, where genomics is not weaponized toward contemporary eugenics, requires that innovation is paralleled by investments in more inclusive social structures that are better prepared for and adaptive to the growing needs of a diverse society.

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