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Effect of Male Partner's Involvement in a Woman's Prenatal Decision-Making Process

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ABSTRACT OF THE THESIS

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By

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Master of Science in Genetic Counseling

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Professor Manuel Porto, M.D., Chair

During the prenatal genetic counseling session, women are given various genetic testing options to evaluate the health of their pregnancy and often seek advice from their male partner whether testing, and which test, should be chosen. Past studies have examined how women make prenatal genetic testing decisions, but little has been studied regarding how the male partner assesses prenatal genetic testing, or the effect of the male partner's involvement in genetic testing decisions. 30 women who came alone and 55 couples at two sites were surveyed following their genetic counseling appointment regarding their genetic testing decisions and were asked to rank the importance of various factors affecting their decisions. Women alone were significantly less likely to be married, more likely to act as the primary decision-maker during pregnancy and reported that their male partner's impact on the genetic testing decision was lower. According to this study, women presenting for prenatal genetic counseling alone, regardless of why they came alone, are more likely to be confident making decisions independently of their partners. Therefore, prenatal genetic counselors should focus on how these women make decisions

autonomously. Women presenting with their partners will rely more heavily on a shared decision-making model, incorporating their male partner's concerns. Within these couples, men are most concerned about the cost of testing or insurance worries, while women are more likely to be concerned about the pain or risk of any invasive testing. Prenatal genetic counselors can use these factors to direct shared decision-making between the couple.

INTRODUCTION

1. Basis of Prenatal Screening/Testing

Prenatal screening and testing offers couples the ability to detect various chromosomal disorders, single-gene disorders, birth defects, and other genetic conditions during a pregnancy. There are a variety of screening and diagnostic tests available to detect these conditions at distinct stages of the pregnancy. Each available test can detect different conditions with different sensitivities and specificities. Diagnostic procedures, such as an amniocentesis or chorionic villus sampling (CVS), can be used to collect fetal or placental tissue for a variety of tests, with near 100% sensitivity and specificity for complete chromosome analysis.[1] The sensitivity and specificity for genetic studies, aside from chromosome analysis, will depend on the type of test and laboratory, and thus, will not always be near 100%.[1] Screening tests, such as maternal serum screening and Non-Invasive Prenatal Screening (NIPS), offer a non-invasive method to detect certain chromosome conditions, such as aneuploidies, and more recently, certain chromosomal microdeletion and microduplication syndromes. An abnormal number of chromosomes is called an aneuploidy. A trisomy is a type of aneuploidy where there is an extra copy of that chromosome, instead of the usual two copies. Trisomy 21 (also known as Down syndrome), trisomy 18 and trisomy 13 are the common aneuploidies that screening and diagnostic tests seek to detect in pregnancy. Screening tests cannot diagnose a condition but can offer a calculated probability for certain conditions.

Before either of these screening tests was available, only invasive diagnostic tests and ultrasound screening options for women to detect or screen for these chromosome conditions.[2] The introduction of these screening tests created a variety of different testing options available for couples to evaluate aneuploidy risk. When paired with routine ultrasound procedures, many birth defects, chromosomal disorders (mainly aneuploidies), and other genetic conditions (such as Smith-Lemli-Opitz syndrome) can be detected without any invasive procedures. As such, an increasing number of couples feel comfortable with just screening tests and ultrasound examinations during the pregnancy.[3,4] Detection of these conditions does not equate to a diagnosis though, as these are merely screening tests. Therefore, diagnostic procedures remain a staple of prenatal diagnostics as the only option with near 100% sensitivity for complete chromosome analysis, and with varying sensitivity for other genetic conditions.[1,2]

2. Prenatal Screening/Testing Options

Historically, couples have had two different choices for diagnostic testing: CVS and amniocentesis. Chorionic villus sampling (CVS) is a procedure that collects placental cells and is typically performed in the 1st trimester between the 10th and 13th week of gestation.[1,5] The amniocentesis is another procedure that collects amniocytes and is typically performed in the 2nd trimester between the 15th and 20th week of gestation.[1,5,6] Both procedures allow couples to have a highly definitive and accurate genetic study performed. Usually, these procedures are performed for chromosome analysis with a karyotype or microarray, but many other genetic tests can be performed on the collected

tissue. The accuracy of chromosome analysis following either procedure is nearly 100%, and thus offers a highly definitive option for couples.[5,6,7]

Back in the 1930s, the use of transabdominal amniocenteses for prenatal health testing was initially used in management of Rh immunization.[8] A couple decades later, amniocentesis as a method for isolating fetal amniocytes, began to gain widespread acceptance as a method for detecting numerous chromosomal and other genetic conditions.[7] In 1972, amniocentesis was enhanced with ultrasound guidance to help determine puncture site and reduce risk to mother and fetus. As the various genetic testing modalities expanded, the amniocentesis procedure allowed for amniocytes to be cultured for additional genetic studies using these new modalities.[6] This procedure can now be utilized to detect even smaller microdeletions and microduplications of the chromosomes. As amniocentesis utilization increased, guidelines for implementation were developed with regular adjustments to fit the ever-changing clinical landscape.[6,9]

In 1960, trans-cervical chorionic villus sampling (CVS) was introduced but was not utilized commonly until the methodology later improved in the 1980s.[10] By 1984, the trans-abdominal technique was introduced and made available for prenatal diagnosis.[10] Both procedures were utilized to extract tissue from the chorion for genetic testing studies, such as a karyotype for aneuploidy detection. Other single-gene tests were introduced starting in the 1990s, such as for Cystic Fibrosis.[1,6,7] Extensive training and practice is required of any physician to perform either the trans-cervical CVS or the trans-abdominal CVS.[1,2]

Even though both CVS and amniocentesis remain nearly 100% sensitive for chromosome analysis of aneuploidies, couples are often leery of the two procedures (CVS

and amniocentesis) due to a variety of reasons, one of which is the procedure-related risk of miscarriage. With every amniocentesis or CVS, there is a small procedure-related risk of miscarriage. Studies have shown that even when performed by a trained professional, the risk of miscarriage associated with amniocentesis ranges from 0.11% to 0.50%, with the risk of miscarriage from CVS procedures at approximately the same rate.[5,11] Some more recent studies report that the risk from either test could be slightly lower when performed at experienced centers.[11,12] These risks present an important psychosocial counseling dilemma, since some couples will not chance that risk under any circumstances and other couples will wish to proceed with the highly accurate and definitive test over the screening options.

Following the introduction of the CVS and amniocentesis, prenatal genetic screening was introduced in the 1970s as elevated levels of maternal serum alpha fetoprotein (MSAFP) in the second trimester were seen more commonly in fetuses affected with open neural tube or abdominal wall defects.[13] This finding sparked the development of further maternal serum screening tests in the second trimester of pregnancies. In 1984, MSAFP was found to be significantly lower in fetuses with trisomy 21 compared to unaffected pregnancies.[14] At this point, MSAFP screening in the second trimester was recognized as an efficient and cost-effective method for detecting trisomy 21, neural tube and abdominal wall defects.[13,15] Then, in 1988, pregnancies affected with trisomy 21 were shown to have significantly lower levels of unconjugated estriol (uE3) and elevated levels of human chorionic gonadotropin (hCG).[16,17] When combined, MSAFP, uE3 and hCG,, known as the "triple screen," became the first three biochemical markers utilized in trisomy 21 screening. In 1993, the association of trisomy 18 with decreased levels of MSAFP, hCG, and

uE3 helped begin the screening for trisomy 18.[18] Shortly afterwards, levels of inhibin-A in maternal serum were found be significantly increased in fetuses affected with trisomy 21, leading to the fourth analyte added to the screen, now called the "quadruple screen". Further associations were then found with these markers and trisomy 13.[19] While several laboratories currently use these markers to screen for trisomy 13, ACMG published a statement in 2009 recommending not to screen for trisomy 13 with this method due to low detection rates.[19] Further analysis of pregnancies with drastically low uE3 values opened the doorway for screening for a rare genetic condition called Smith-Lemli-Opitz syndrome.[20] These extremely low uE3 values also coincided with increased chance of fetal demise or other adverse pregnancy outcomes.[20]

Prenatal biochemical marker screening continued to evolve with the discovery of associations of elevated free β-hCG and low levels of PAPP-A with trisomy 21 in the first trimester of pregnancy.[21,22] Later, it was found that pregnancies affected with trisomy 18 and trisomy 13 also showed markedly low levels of PAPP-A, but unlike trisomy 21 had lower levels of free β-hCG.[24,25] This allowed for detection of trisomy 18 and trisomy 13 along with trisomy 21 using the same biochemical markers.[24,25] In addition, increased fetal nuchal translucency thickness in the first trimester of pregnancy was found to be strongly correlated with chromosomal abnormalities.[23] It was then deduced that combining first trimester nuchal translucency with PAPP-A and β-hCG would be an efficient and cost-effective screening method for aneuploidy detection in the first trimester.[26]

Pairing of both first trimester and second trimester screening options (also called integrated screening) with maternal age has proven to be an effective method for detection of trisomy 18 and trisomy 21.[27] It was initially described that integrated screening can

detect 85% of pregnancies affected with trisomy 21.[27] More recently, detection rates of 90% and higher have been reported for trisomy 21, and around 80% for trisomy 18 and 13, depending on the methodology.[28,29,30] The integration of these screening methods together has shown to have higher detection rates than utilizing maternal age alone.[26,27,28] These maternal screening methods are a staple of routine aneuploidy screening and have assisted in the detection of up to 80% to 90% of pregnancies affected with these aneuploidies.[28,29,30]

For the couples who are wary of procedure-related risks of diagnostic testing, non-invasive screening tests allow for accurate aneuploidy risk assessment without an increased risk of losing the pregnancy. In these cases, maternal serum screening that utilizes first and second trimester screening with a nuchal translucency ultrasound scan is a great source of information and, at times, relief for many couples concerned about aneuploidy in their pregnancy.[26,27]

After years of implementing maternal serum screening to detect aneuploidy, the discovery of circulating cell-free fetal DNA in maternal plasma initiated the introduction of a new prenatal screening test, called cell-free fetal DNA screening or Non-Invasive Prenatal Screening (NIPS).[31] It was determined that in maternal serum, the circulating fetal DNA was sufficient for reliable detection of some aneuploidies. Chromosomal dosage could be quantified in the circulating DNA to determine risk for some aneuploidies. In 2011, Sequenom Center for Molecular Medicine started offering NIPS as a highly accurate, non-invasive method for detecting chromosomal aneuploidy in high-risk pregnancies.[31] Compared to biochemical screening, ultrasound, and maternal age, NIPS has much higher detection rates, sensitivity, and specificity for trisomies 13, 18, and 21 than routine

maternal serum secreening.[32,33,34] Following meta-analysis of several studies, sensitivities for trisomy 21 are reported at approximately 99.8%, trisomy 18 at approximately 97.7%, trisomy 13 at approximately 91.7% and a range between 70% and 95% for sex chromosome aneuploidies (SCAs).[32,33,34] The reported specificities for trisomy 21, 18 and 13 is all approximately 99% making it an ideal screening test at limiting false negative results.[32,33,35] SCAs have a reported specificity as high as 99.6%.[35] Aside from the high sensitivity and specificity, the positive-predictive value (PPV), or the likelihood of a positive result being a true positive, and the negative-predictive value (NPV), the likelihood of a negative result being a true negative, are excellent indicators of the accuracy of a positive or negative result from NIPS. The PPV can be as high as 99% and negative-predictive value (NPV) can be as high as 99.9%. The PPV can be much lower, depending on the a priori risk in the pregnancy.[32,33] The PPV and NPV can be much lower for SCAs and, again, depend highly on the initial risk of the aneuploidy in question.[32,33]

After its introduction, other genetic testing laboratories began to offer their own version of NIPS with slightly different reported sensitivities and specificities.[32] Nonetheless, the evidence shows that NIPS acts as a highly effective tool for detection of aneuploidy in pregnancies.[32,33,34] At its inception, NIPS was recommended to only be offered to those at high risk for aneuploidy.[35] Recently, ACMG has reported that NIPS should be offered to more than just high-risk pregnancies, possibly leading to an expansion in usage of this technology as a population screening tool.[34] The continued evolution of non-invasive prenatal screening (NIPS) has offered a highly accurate, non-invasive screening option for couples who wish to avoid the procedure-related risks of an amniocentesis or CVS.[32,33]

3. Chromosome Conditions & Analysis

Aneuploidies are most often due to a fault in separation of chromosomes during gametogenesis, called nondisjunction, that seemingly occurs sporadically.[37] As women age, the chances of a sporadic trisomy gradually increase, which is the reason why women who are 35 or older at delivery are considered to be at a higher risk for aneuploidies.[37] The incidence of sex chromosome aneuploidies (SCAs), which affect chromosomes X and Y, does not appear to follow this trend with advancing maternal age.[38]

Trisomy 21, more commonly known as Down syndrome, is a chromosomal disorder caused by an additional copy of chromosome 21. Individuals with trisomy 21 can have multiple malformations, congenital anomalies of the heart, characteristic facial features, intellectual impairment and some other common features.[39] The phenotype of individuals with trisomy 21 can vary and the life span is reduced, especially when there are severe heart defects.[39]

Trisomy 18, also known as Edwards syndrome, is caused by an additional copy of chromosome 18. Individuals with trisomy 18 often have multiple congenital abnormalities including defects in the heart and various other organs.[40] Trisomy 18 is associated with severe intellectual disability. Depending on the gestational age of diagnosis, between 48-60% will result in a live birth.[41] Individuals often do not survive past the first few months of life due to the variety of severe anomalies at birth. Only 5% of these individuals will survive for more than a year, with very few surviving into childhood or adolescence.[40]

Trisomy 13, also known as Patau syndrome, is due to an additional copy of chromosome 13. Individuals with trisomy 13 often have several congenital abnormalities in various organ systems, congenital heart defects and severe intellectual disability.[42]

Following the prenatal diagnosis, the survival to live birth can vary with gestational age at diagnosis and range from 30-40%.[41] Only about 50% of live births will survive past the first week of life, with approximately 3-10% not surviving past the first year of life.[42]

Sex chromosome abnormalities (SCAs) are a group of chromosomal disorders characterized by extra or missing copies of the X or Y chromosome. Some of the most common SCAs are Turner syndrome (45, X), Klinefelter syndrome (47, XXY), and triple X syndrome (47, XXX). Each syndrome has characteristic features, but all are considered to be milder than the previously discussed disorders.[43,44] Cognitive ability can vary greatly depending on the type of SCA, with most having normal cognitive function.[43] Each SCA has a distinct phenotype. Commonly, individuals with a diagnosed SCA may be infertile and some have characteristic behavioral features.[44]

In order to detect these chromosomal conditions, a variety of studies are available following either a CVS or amniocentesis. Traditionally, chromosome analysis has always relied upon karyotypes to analyze the full complement of chromosomes for detection of aneuploidy, large deletions and duplications of chromosome material, and large structural chromosome anomalies, such as translocations.[45,46] Karyotype analysis is performed to allow for couples to most accurately determine if their pregnancy has any of these cytogenetic abnormalities. However, in the last couple of years, the rise of chromosomal microarray technology provides an additional option. Chromosomal microarray analysis can improve the detection of smaller copy number variants (CNVs) compared to traditional karyotypes, and is recommended as a first-line test when one or more anomalies are present on ultrasound.[47,48,49] Several studies have shown that the chromosomal microarray has increased detection of cytogenetic abnormalities by 6-7% when an ultrasound anomaly

is present, and by 1.7% for couples referred due to advanced maternal age or a positive serum maker screen with normal ultrasound scans.[47,48] While chromosomal microarray technology has increased detection for cytogenetic abnormalities in pregnancies, it is limited as it cannot detect triploidy, depending on the platform used, or balanced rearrangements, both of which can be detected with a karyotype. In addition, the new technology has revealed more variants of unclear significance (VUS), which are defined as variations in the genetic sequence for which association with disease risk is unknown and, can thus, create a challenging counseling situation.[49,50] Therefore, while chromosome microarray analysis continues to become a more popular option for couples to choose due to increased detection of chromosome abnormalities, karyotype still remains a staple of cytogenetic analysis due to the ability to detect balanced translocations and triploidy.

4. Factors Affecting Couples Decision-Making

In a prenatal genetic counseling session, couples are tasked with making a rather difficult decision regarding the health of their unborn child. According to Lawson and Pierson, reproductive decisions are believed to be premised upon the 'rational choice model,' which views couples as autonomous, sensible, individualistic decision makers with choices in pregnancy.[4,51] Their choice has to balance their own beliefs, convictions and experiences with the goal of having a healthy child. Given that every couple is distinct, there are a variety of different factors that can affect and alter how a couple approaches a genetic testing decision. A recent study looked to examine all the possible factors that influence reproductive decisions and narrowed it down to the following: perceived pain of diagnostic testing, anxiety about health of fetus, emotional and societal burden of possible

termination, perceived risk of diagnostic testing, limitations and uncertainty of genetic tests, financial and social burden of a child with disabilities, access to prenatal services, health care education and support, personal philosophy, past pregnancy history, media, social support, ethnicity, and socioeconomic status.[3,4] Each factor above has been shown to contribute to a woman's decision-making in varying degrees and for various reasons. While the choices that women may make have been extensively analyzed, there is still more to learn about the contributing factors of their male partners.

One of the most prominent factors for a couple's genetic testing decision is the indication for testing. The nature of genetic counseling is to be nondirective and always allow for couples to make a rational decision based upon the information available to them and their own beliefs.[3,51] While genetic counselors always aim to remain nondirective, the indication for testing can skew decision-making for couples. When couples are notified of extensive, serious or complicated prenatal ultrasound findings, screening tests may not be the best option for detection of the root cause for the findings.[52,53] A recent study revealed that 86.7% of people declined an invasive procedure after a normal ultrasound scan.[52] In addition, pre-conceived beliefs strongly determine the ultimate decision even before an ultrasound scan has occurred. Approximately 5.3% of women who had an abnormal ultrasound and 12.9% of women who had a normal ultrasound changed their initial decision about an invasive procedure following genetic counseling.[53,54] When couples present to genetic counseling due to a positive maternal serum screen or advanced maternal age, they may feel more comfortable gathering more information through a screening test, such as NIPS, before proceeding with any invasive procedures.[52,54] The

difficulty in predicting how couples react to these indications is rooted into their own personal beliefs, level of anxiety, and perceptions of the risk in the pregnancy.[3,4,51]

Another central theme to evaluating how a couple makes decisions is their cultural or religious background. Paternalistic cultures and religions focus on a male-centered approach to decision-making, while other cultures may leave all responsibility to the mother as the decision-maker. For example, in a study of Latina women, they were more likely to decline testing compared to the general population possibly, as the authors posited, because they are a part of a more religious, male-dominated, family-centered and possibly superstitious culture.[55] In Asian cultures, women were actually found to be more favorable towards genetic testing, along with white women, even if they were less informed about the test itself.[56] Several cultures focus on very traditional values that may be against the option of terminating a pregnancy. Due to these values, many couples may feel uncomfortable discussing invasive testing because it may be viewed as useful only if considering pregnancy termination of an affected pregnancy. In fact, they may even be averse to some screening tests because of the possibility of a positive screen leading to invasive testing or a discussion of pregnancy termination. In these cases, it is vital to have an upfront discussion with the couple about their plan if any abnormal results should return.

In addition to some cultural beliefs, there are many religions that have stances against pregnancy termination. Some couples see invasive procedures as a pathway that leads toward termination in the event of an abnormal result. For example, in the Islamic faith, termination is allowed before the 40th day after conception, and in some medically-significant cases, before the 120th day after conception.[57] For this reason, many religious

couples may shy away from invasive procedures and may not want to pursue any testing at all. Although these themes tend to drive the decision-making agenda for some couples, there are studies that show conflicting views, in which religion and race did not significantly alter prenatal genetic testing decisions.[58] So, it is best to approach couples and help them guide their own decision without assumptions about what that decision may be.

The ability to guide couples through the shared decision-making process is one of the many competencies of a genetic counselor.[51,59] Genetic counselors foster a discussion that encompasses the beliefs and goals of the couple in hopes of guiding them to make a genetic testing decision that is best for the couple.[51,59] Genetic counselors can utilize their expertise in genetics to explain challenging concepts and lead into a discussion about what test would be best for the couple.[51,59] Since genetic testing is often a very personal decision, genetic counselors must remain non-directive to avoid influencing the decision. Non-directiveness is an approach to providing genetic information without influencing the decision by fostering discussion. In this way, genetic counselors do not often affect the genetic testing decision made, in contrast to physicians who may not ascribe to a non-directive approach.[3,59] So, while genetic counselors may interact heavily with the patients, they seek to utilize their skills to foster a shared discussion between the couple and avoid directly influencing the decision.

5. The Role of the Father

From a historical perspective, women have been viewed as the child bearer and caregiver.[60,61] So, the reproductive decisions and responsibilities that accompany pregnancy have long fallen into their role.[60,61] Men have always been encouraged to

participate in the pregnancy but have always been seen primarily as supporters to the women throughout pregnancy. Men's role as supporters and protectors of the women places them as secondary members in the couple, and thus, secondary decision makers.[60,61] For instance, in the past, men were not even allowed into the delivery room. In the past several years, there has been a surge promoted by the World Health Organization (WHO) to increase paternal involvement in reproductive decisions and pregnancy.[62] In fact, this push has stemmed partially from the fact that men do not feel included in the decisions made during pregnancy, despite all efforts to make them feel included.[60,61,62,63] They often feel as if they receive information from these appointments secondhand, and have noted that clinicians will often focus more on the patient (the pregnant woman) than themselves.[62,64,65] While male involvement has vastly improved over time, there are still gaps in research and information on the male preference in regards to reproductive decisions.

Recent studies have brought up four major themes that male partners feel are a barrier to their inclusion in prenatal screening and testing.[60,61,62] These barriers include: ambivalence, the wave of emotions, shared decision-making, and limited engagement with health care professionals.[62,65] While more research is needed to analyze and determine further barriers, these are the most commonly recognized at this point. The barrier for ambivalence stems from the doubt or uncertainty regarding medical risk and uncertainty with the large amount of medical information. They felt that the extra worry of screening was unnecessary and questioned the accuracy of screening tests. They were also worried about being swept along on the testing cascade. This barrier works closely with a lot of the emotional barriers for men.[62,65] Men have consistently been thought of as the supportive

partner who does not get upset or emotional, even if they have strong emotions about the subject matter.[60,63,66] So, when the majority of emotional support is directed at the woman, the man may not feel adequately supported himself. In addition, men feel a strong need for a shared decision-making model for all reproductive decisions.[60,61] While the woman may be the primary decision-maker in reproductive decisions, men may wish to be included in the process, and thus, contribute to the shared decision. This lack of shared-decision making acts as a barrier for men to feel integrally involved in the process.[62,65,66] Finally, the lack of engagement with health care professionals is a barrier for them because, while the woman may be the official patient, genetic screening and testing affects the status of their pregnancy as well. They wish to feel as if any decisions are made as a unit and that the health care professionals see the couple as a unit, rather than seeing the partner as just a support system.[62,65] In conclusion, as men are encouraged to increase involvement in all aspects of reproductive decisions and prenatal genetic testing, it is important to support their involvement and help foster a shared-decision making process.

6. Hypothesis

The goal of prenatal genetic testing and screening is to provide couples with information about genetic risk and to discuss the health of their pregnancy. Oftentimes, a variety of different genetic testing options for detection of a variety of different chromosomal disorders and other genetic abnormalities are additionally discussed. As the number of testing options continues to increase, decisions about what, if any, testing to undergo becomes much more challenging for couples to make.[6,22] Expectant mothers are looking for guidance and assistance in making this decision.[3,4,53] Non-directiveness remains

central to genetic counseling, so pregnant women will seek assistance from their male partners, family, friends and other support systems in making decisions regarding genetic testing. So far, little data has been collected to further our understanding of how male partners impact this decision.[54,57,58] Clearly, it is imperative for us to better understand what effect the male partners have on the decision-making process and to further evaluate the shared-decision making dynamic. This study will aim to understand how the presence or absence of the male partner in the genetic counseling session impacts this genetic testing decision. Based on prior research and observation, it is expected that women will feel more comfortable making invasive testing decisions with their partner present and more likely to choose no testing at all with their partner absent. In addition, women who come with their partner will favor engaging in a shared decision-making model while women without their partner will feel more confident making testing decisions alone.

MATERIALS & METHODS

1. IRB

This study was reviewed under exempt status by the Institutional Review Board (IRB) at the University of California, Irvine (UCI) (IRB# HS 2017-3913) and at the St. Jude Medical Center (SJMC) (IRB# 18-011). SJMC was included as an off-site location for data collection underneath the UCI IRB following approval from both institutions.

2. Participants/Recruitment

Participation in the study was restricted to literate men and women over 18 years of age who spoke English or Spanish. Following prenatal genetic counseling for a variety of referral types, women who came alone and couples were offered the opportunity to participate in the research study through a brief survey. Women who attended their genetic counseling session alone took a survey that is distinct from the surveys that were given to each couple that presented to genetic counseling. Within each couple, both the woman and her partner were given separate surveys to be filled out on their own. All questions across each of the three surveys were standardized with minor adjustments to the wording in order to be directed to the participant filling out the survey.

Recruitment began in January of 2018 at the Center for Fetal Evaluation at the University of California, Irvine Medical Center in Orange, CA. In March of 2018, recruitment was initiated at SJMC once IRB approval was granted. Participants were recruited for an optional, anonymous survey following their initial genetic counseling session in the prenatal clinic at either UCIMC or SJMC. If the participant wished to learn more, the genetic

counselor or lead researcher reviewed the project and answered all follow-up questions. Oral consent was obtained by the genetic counselor using a verified recruitment script in either English or Spanish, depending on the preferred language of the participant. All recruiters were competent in both English and Spanish. Each participant was then given a Study Information Sheet in their language of choice with the contact information of the lead and co-researcher of the study. Afterwards, each participant was given their own paper questionnaire in either English or Spanish to be filled out in the private waiting room outside of their appointment. Participants were given instructions to place their finished questionnaires into a locked drop box in the waiting room for which only the lead and co-researchers had access to upon completion.

On a weekly basis, the drop boxes were emptied, the results of which were recorded in a Microsoft Excel spreadsheet. The data was then entered into Statistical Package of the Social Sciences (SPSS), a statistical analysis software for logical batched and non-batched analysis.

3. Survey Descriptions

Three separate questionnaires were designed for this study: one for women who attended their appointments alone, one for women who attended with their partners and one for their partners. All questionnaires were designed by the lead researcher with assistance from the co-researchers and all questions were original or adapted from previous psychometrically-analyzed questionnaires. All questionnaires were designed in English and then translated into Spanish. Every questionnaire was given a distinct identifying code and no identifying information was gathered from the participants. In this

way, each questionnaire could be uniquely distinguished while remaining anonymous. In addition, the questionnaire for each woman could be linked to their partner's questionnaire for matched-analyses.

Each questionnaire contained a series of questions directing the participant through common demographic questions, questions about their testing choice, what factors affected that testing choice and questions that were aimed at determining how the couple made decisions. The goal of the questions was to elucidate the exact factors that affected the genetic testing decision of women who came alone and those who came with their partner. These factors could then be used to compare the two groups of women to further understand how women make decisions with and without their partners. In addition, a questionnaire was given to each partner in order to determine what factors were important to them with regard to making decisions about prenatal genetic testing.

4. Statistical Analysis

Analyses were completed using IBM SPSS Statistics version 25. Primary comparisons were women who came alone versus who came with their partner with both institutions (UCIMC and SJMC) pooled. Data was compared between the three major groups (women who came alone versus women who came with their partner). Major categorical variables in the demographic section (ethnicity, age, relationship status, pregnancy count and number of children) were analyzed using a Pearson Chi-Squared test of independence to examine for any significant differences between the groups. A Pearson Chi-Squared test of independence was used to detect for any differences between the different groups of women with regard to referral indication or genetic testing decisions as well. A Mann-

Whitney U test analyzed the ordinally ranked factors regarding each group's testing decision. For women who came with their partner and their linked male partner, a Wilcoxon Matched-Pairs Signed-Rank test was utilized to analyze for significant differences within each couple's factors, to further determine male versus female preferences within each couple. Finally, the relationship dynamic questions were analyzed for significance using a Mann-Whitney U test.

RESULTS

1. Demographics

A total of 140 of participants were recruited from University of California, Irvine Medical Center (UCIMC) and St. Jude Medical Center (SJMC) between January 2018 and April 2018. Participants were divided into three groups: women who came alone, women who came with their partner, and the male partners who accompanied their female partner to the genetic counseling session. Demographic information such as age group, ethnicity, education level, pregnancy count, number of children and relationship status were collected for each of these groups (Table 1). In addition, the women who came alone self-reported additional demographic questions regarding their male partner who could not attend the session. The most frequently reported age group, ethnicity, relationship status and education level were 36-45 (44%), Hispanic/Latino (38%), married (78%) and college degree (32%), respectively. 37% of women reported 3 or more pregnancies, while 38% of respondents reported having 0 children. The majority (65%) of participants were recruited from SJMC.

Demographic Info	Women who came alone (n=30)	Partners of Women who came Alone (n=30)	Women who came with partner (n=55)	Partners who came to appointment (n=55)	Total of all 4 groups (n=170)
Age Group					
Under 25	2 (7%)	2 (7%)	4 (7%)	3 (5%)	11 (6%)
25-35	14 (47%)	12 (40%)	29 (53%)	25 (45%)	70 (41%)
36-45	14 (47%)	15 (50%)	21 (38%)	24 (44%)	74 (44%)
46-55	0 (0%)	1 (3%)	1 (2%)	3 (5%)	5 (3%)
Ethnicity					
African American/Black	0 (0%)	1 (3%)	0 (0%)	1 (2%)	2 (1%)
White/Caucasian	9 (30%)	9 (30%)	19 (35%)	23 (42%)	60 (35%)
American Indian/Alaskan Native	0 (0%)	0 (0%)	0 (0%)	1 (2%)	1 (1%)
Hispanic/Latino	15 (50%)	15 (50%)	17 (31%)	17 (31%)	64 (38%)
Asian/Pacific Islander	6 (20%)	5 (17%)	18 (33%)	13 (24%)	42 (25%)
Other	0 (0%)	0 (0%)	1 (2%)	0 (0%)	1 (1%)
Education					
Less than a high school degree	2 (7%)	5 (17%)	1 (2%)	0 (0%)	8 (5%)
High school diploma	7 (23%)	8 (27%)	9 (16%)	11 (20%)	35 (21%)
Some college	6 (20%)	8 (27%)	12 (22%)	11 (20%)	37 (22%)
College degree	9 (30%)	7 (23%)	20 (36%)	18 (33%)	54 (32%)
Postgraduate degree or higher	6 (20%)	2 (7%)	13 (24%)	15 (27%)	36 (22%)

Table 1. Frequencies and proportions of demographical information

Demographic Info	Women who came alone (n=30)	Partners of women who came alone (n=30)	Women who came with partner (n=55)	Partners who came to appointment (n=55)	Totals
Number of children					**Total (n=140)**
0	10 (33%)		21 (38%)	22 (40%)	53 (38%)
1	11 (37%)		21 (38%)	18 (33%)	50 (36%)
2	7 (23%)		9 (16%)	10 (18%)	26 (19%)
3+	2 (7%)		4 (7%)	5 (9%)	11 (8%)
Number of pregnancies					**Total (n=85)**
0	6 (20%)		14 (26%)		20 (24%)
1	6 (20%)		12 (22%)		18 (21%)
2	6 (20%)		10 (18%)		16 (19%)
3+	12 (40%)		19 (35%)		31 (37%)
Relationship Status					**Total (n=140)**
Single	4 (13%)		2 (4%)	1 (2%)	7 (5%)
Widowed	0 (0%)		0 (0%)	0 (0%)	0 (0%)
Committed relationship	8 (27%)		7 (13%)	9 (16%)	24 (17%)
Married	18 (60%)		46 (84%)	45 (82%)	109 (78%)
Divorced	0 (0%)		0 (0%)	0 (0%)	0 (0%)
Separated	0 (0%)		0 (0%)	0 (0%)	0 (0%)
Collection Site					**Total (n=140)**
UCI	17 (57%)		16 (29%)	16 (29%)	49 (35%)
SJMC	13 (43%)		39 (71%)	39 (71%)	91 (65%)

Table 1 (continued). Frequencies and proportions of demographical information

Demographic information was grouped to increase frequencies and perform a two-sided ($H_0 \neq H_a$) Pearson's Chi-Squared test of independence with a significance level (α) cut-off of 0.05 (Table 2 through Table 8). Ages were grouped for each of the four groups (women who came alone, partners of women who came alone, women with partner and partners who attended the session) by those that are under the age of 36 and those that are

age 36 and over. No statistically significant difference in age groups was detected between the two groups of women (p=0.552) or the two groups of partners (p=0.709) (Table 2).

	Women who came alone (n=30)		Partners of Women who came Alone (n=30)		Women who came with partner (n=55)		Partners who came to appointment (n=55)	
Age Group	**N**	**%**	**N**	**%**	**N**	**%**	**N**	**%**
Under 36	16	53%	14	47%	33	60%	28	51%
36 & Over	14	47%	16	53%	22	40%	27	49%

Table 2. Age groupings distributed by participant type

Ethnicities were grouped to increase power into the following: Hispanic/Latino, White/Caucasian, and "other" (Asian/Pacific Islander, African American/Black, American Indian/Alaskan Native and all others). No significant difference (p-value = 0.183) was detected between the three ethnic groupings across the two groups of women (p=0.183) or their partners (p=0.221). Although, the "other" group was noticeably higher (35%) in the women who came with partner group. In addition, for women who came alone, there was a noticeably higher proportion of both women (50%) and their partners (50%) who identified as Hispanic/Latino. Both the White/Caucasian group (42%) and the Asian/Pacific Islander group (27%) were higher in the partners who came to the appointment.

	Women who came alone (n=30)		Partners of Women who came Alone (n=30)		Women who came with partner (n=55)		Partners who came to appointment (n=55)	
Ethnicity	**N**	**%**	**N**	**%**	**N**	**%**	**N**	**%**
Hispanic/Latino	15	50%	15%	50%	17%	31%	17%	31%
White/Caucasian	9	30%	9%	30%	19%	35%	23%	42%
Asian/Pacific Islander and All Others	6	20%	6%	20%	19%	35%	15%	27%

Table 3. Ethnicity groupings among different participant type

Education level was categorized to increase power into those with a college degree or higher and those with less than a college degree (Table 4). No significant difference (p-value = 0.374) in education level was detected in education between the two groups of women. The male partners' education levels were significantly different as 70% of those who did not attend the appointment had less than a college degree compared to only 40% of those who did attend (p-value = 0.008).

	Women who came alone (n=30)		**Partners of Women who came Alone (n=30)**		**Women who came with partner (n=55)**		**Partners who came to appointment (n=55)**	
Education Level	**N**	**%**	**N**	**%**	**N**	**%**	**N**	**%**
Less than a college degree	15	50%	21	70%	22	40%	22	40%
College degree or higher	15	50%	9	30%	33	60%	33	60%

Table 4. Education groupings among different participant type

Number of prior pregnancies was grouped into women with 1 or less and those with 2 or more to increase statistical power (Table 5). No significant difference (p-value = 0.519) was detected between the two groups of women with regard to prior pregnancy count.

	Women who came alone (n=30)		**Women who came with partner (n=55)**	
Number of prior pregnancies	**N**	**%**	**N**	**%**
0 or 1	12	40%	26	47%
2 or more	18	60%	29	53%

Table 5. Number of past pregnancies grouped between both groups of women

Number of children was grouped based on women who had 1 or less children and those with 2 or more children (Table 6). No significant difference (p-value = 0.522) was detected in regard to number of current children.

	Women who came alone (n=30)		Women who came with partner (n=55)	
Number of children	**N**	**%**	**N**	**%**
0 or 1	21	70%	42	76%
2 or more	9	30%	13	24%

Table 6. Number of children grouped between both groups of women

Women were then grouped based off their marital status (Table 7). Statistical significance was detected with regard to marital status as 84% of women with partner were married, compared to 60% of women who came alone (p-value = 0.016).

	Women who came alone (n=30)		Women who came with partner (n=55)	
Marital status	N	%	N	%
Not married	12	40%	9	16%
Married	18	60%	46	84%

Table 7. Relationship/marital status grouped between both groups of women

Table 8 shows the distribution of recruitment sites (UCI and SJMC) between the two groups of women. Statistical significance (p-value = 0.013) was detected with regard to recruitment site with 57% of women who came alone seen at UCI and only 29% of women who came with partners seen at UCI.

	Women who came alone (n=30)		Women who came with partner (n=55)	
Recruitment site	N	%	N	%
UCI	17	57%	16	29%
SJMC	13	43%	39	71%

Table 8. Recruitment site grouped between both groups of women

2. Referral Indication & Genetic Testing Decisions

Table 9 below lists the reasons for referral to a genetic counselor. A Pearson's Chi-Squared test of independence was used to determine statistical significance between the two groups using a two-tailed hypothesis ($H_0 \neq H_a$). No significant differences were noted between the two groups (p-value = 0.842). The most common referral reason was advanced maternal age (60%). Both the positive serum screen and "other" referral reason each represented 20% of all referrals. The "other" referral reason included a write-in option in which primarily included ultrasound anomalies, family history of genetic disorders, pregnancy history or teratogen exposures.

	Women Alone (n=30)		**Women with Partner (n=55)**		**Total (n=85)**	
Referral Indication	N	%	N	%	N	%
Advanced maternal age	17	57%	34	62%	51	60%
Positive serum screen	7	23%	10	18%	17	20%
Other (ultrasound anomaly, family history, etc.)	6	20%	11	20%	17	20%

Table 9. Frequencies and proportions of referral indication to prenatal genetic counseling

Table 10 shows the frequency count and proportions for whether the women elected genetic testing or declined genetic testing. A Pearson's Chi-Squared test of independence was used to determine statistical significance between the two groups using a two-tailed hypothesis ($H_0 \neq H_a$). Statistical significance was not detected between the two groups (p = 0.810).

	Women Alone (n=30)		**Women with Partner (n=55)**		**Total (n=85)**	
Genetic Testing Decision	N	%	N	%	N	%
Elected genetic testing	15	50%	29	53%	44	52%
Declined genetic testing	15	50%	26	47%	41	48%

Table 10. Frequencies and proportions of testing decision for both groups of women

Women from either group who elected to proceed with genetic testing were asked to mark the tests they chose. The invasive tests (amniocentesis or CVS with chromosome analysis) were combined in order to increase power for statistical analysis into the invasive testing section (Table 11). The "other" genetic testing choice allowed for a write-in section which most commonly included carrier screening or other parental testing. A Fisher's exact test of independence was used to determine statistical significance between the two groups using a two-tailed hypothesis ($H_0 \neq H_a$). Statistical significance was not detected between the two groups (p-value = 0.977).

	Women Alone (n=15)		**Women with Partner (n=29)**	
Test Type	N	%	N	%
Invasive Testing	1	7%	2	7%
NIPS	14	93%	27	93%

Table 11. Frequencies and proportions of type of test chosen by both groups of women, if genetic testing was elected

Tables 12 and 13 list the frequencies and proportions for each genetic test that would have been elected based on level of comfort with or without partner present. Respondents were allowed to select as many answers as they wished. Table 12 shows responses to a question that was exclusive to women who came alone and declined all testing (n= 15). They were asked on the survey which genetic test they would have been comfortable with, if their partner was present at the session. Table 13 shows responses to a question for the women who came with their partners and elected any genetic testing option (n= 29). They were asked which genetic test would have been elected if their partner was not present at the session. For women who came alone and declined testing, 33% of them would have been comfortable having an invasive test if their partner was

present. For women who came with their partner and elected any testing option, only 13.8% of them would have been comfortable with an invasive testing option if their partner was not present.

	Test comfortable with if partner present (n=15)	
Test Type	N	%
Invasive test with chromosome analysis	5	33.3%
NIPS	7	46.7%
No testing	8	53.3%

Table 12. Testing women alone would have been comfortable with if partner was present

	Testing comfortable with if partner was not present (n=29)	
Test Type	N	%
Invasive test with chromosome analysis	4	13.8%
NIPS	23	79.3%
No testing	6	20.7%

Table 13. Testing women who came with their partner would have been comfortable with if their partner was not present

3. Factors Influencing Testing Decision

Both groups of women and the male partners who came to the session were asked to give rankings (1 being the lowest rank and 5 being the highest rank) for how each listed factor influenced their genetic testing choice. Women who came alone, women who came with their partners and their male partners each ranked the factors separately and independently from each other. The distribution of ranked factors for each given factor across the three groups is listed in the bar graphs below. Graphs 1 through 9 show the proportions for each ranked value (1 through 5) based off the different given factors.

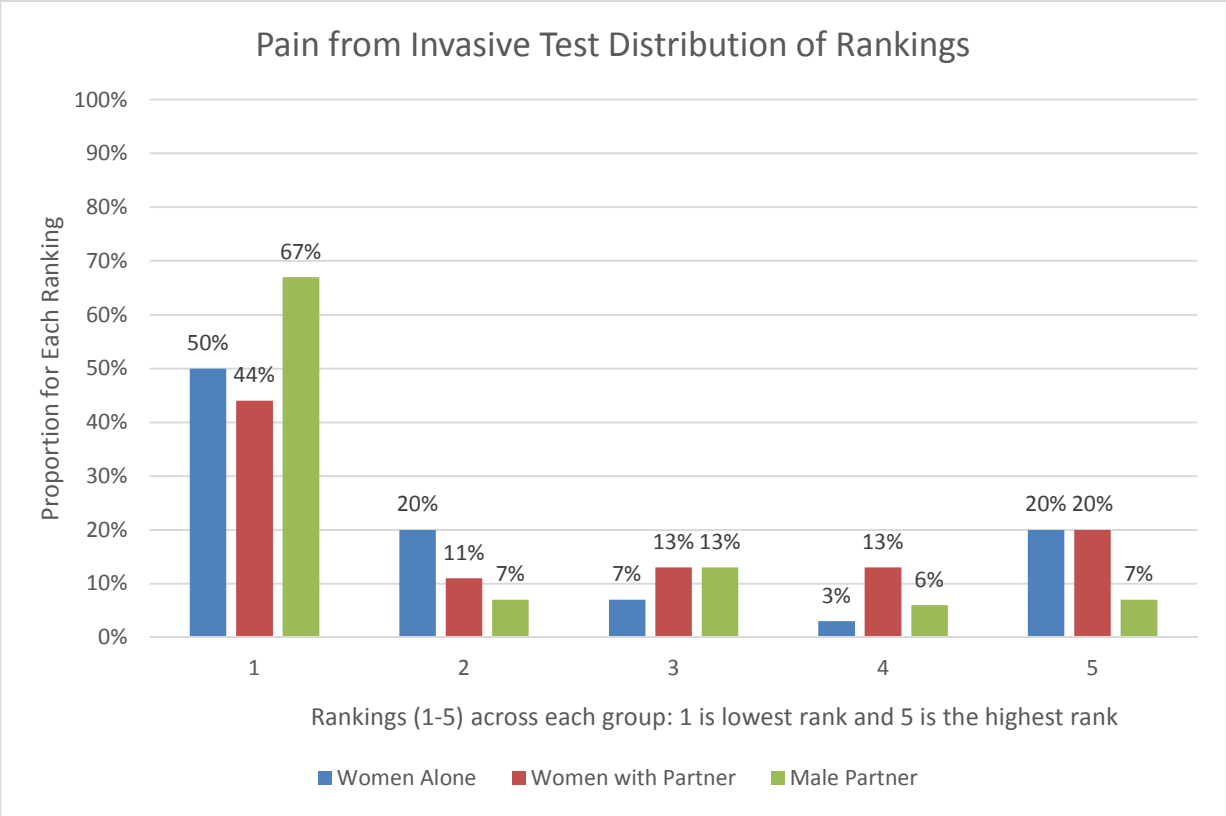

Figure 1. Proportions of each individual and their rankings (1 through 5) on the impact that pain from invasive testing had on their genetic testing decision

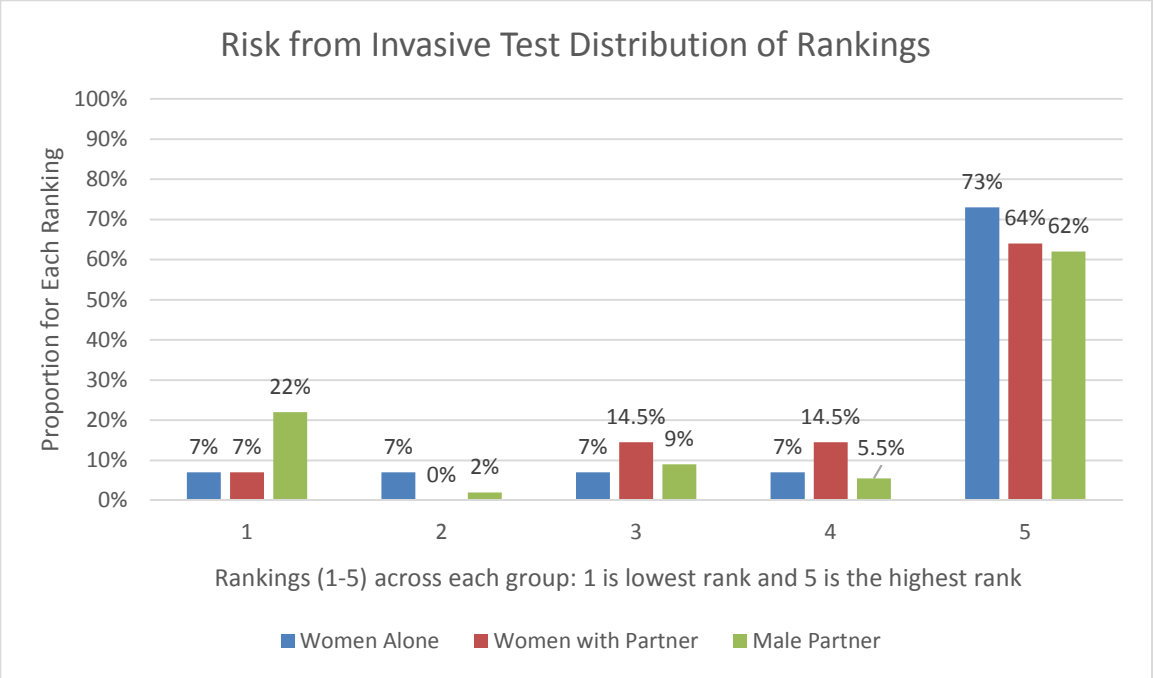

Figure 2. Proportions of each individual and their rankings (1 through 5) on the impact that perceived risk from invasive testing had on their genetic testing decision

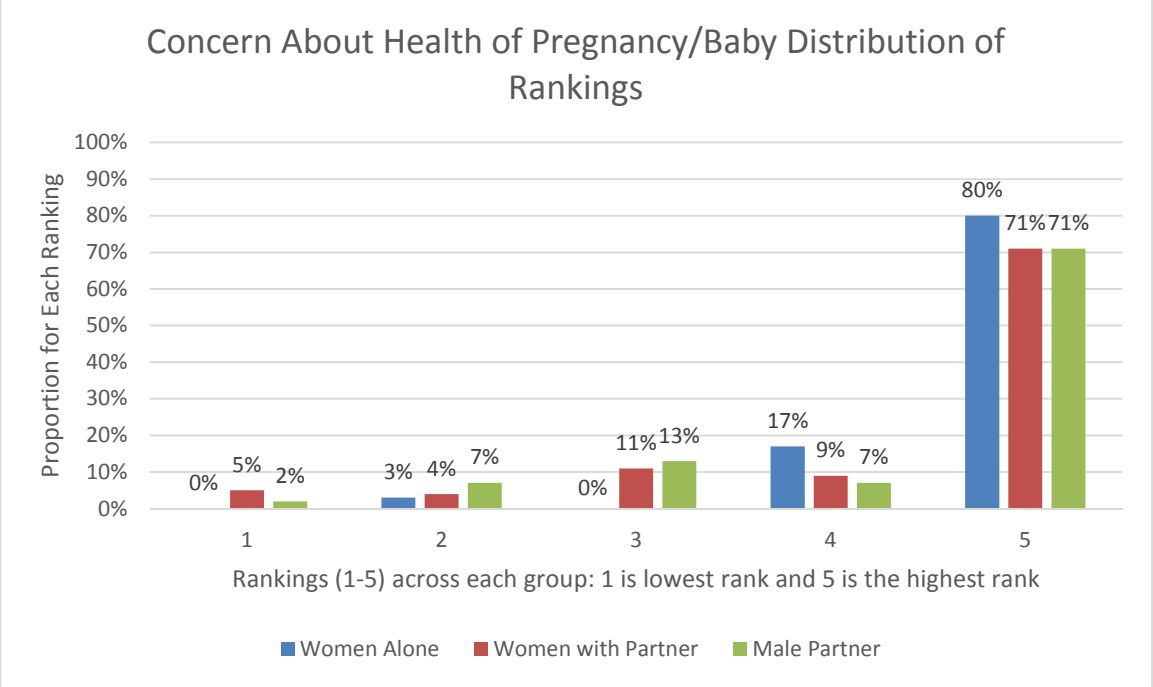

Figure 3. Proportions of each individual and their rankings (1 through 5) on the impact that the concern about the health of pregnancy or baby had on their genetic testing decision

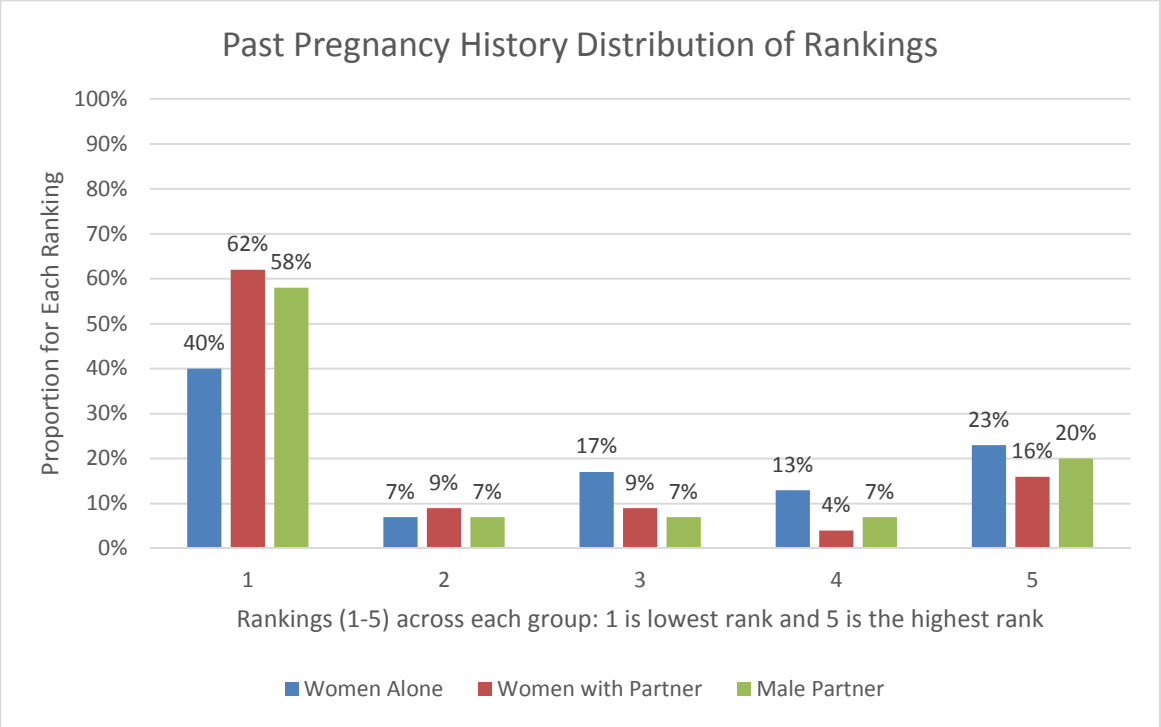

Figure 4. Proportions of each individual and their rankings (1 through 5) on the impact that past pregnancy history had on their genetic testing decision

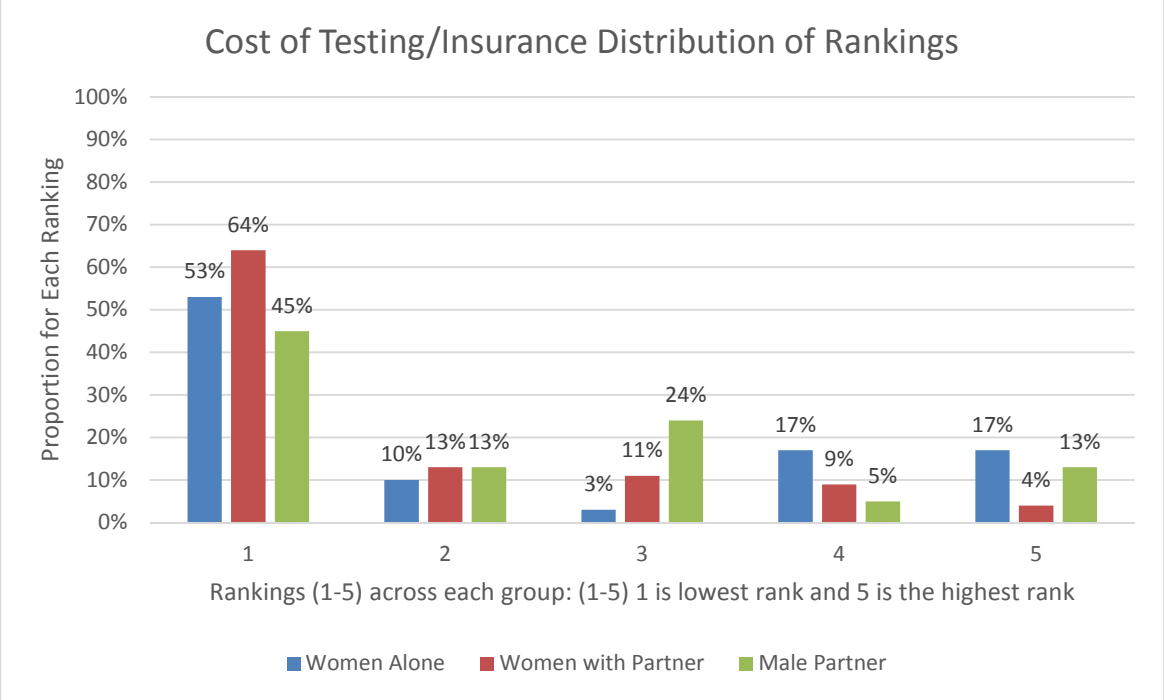

Figure 5. Proportions of each individual and their rankings (1 through 5) on the impact that cost of testing or insurance worries had on their genetic testing decision

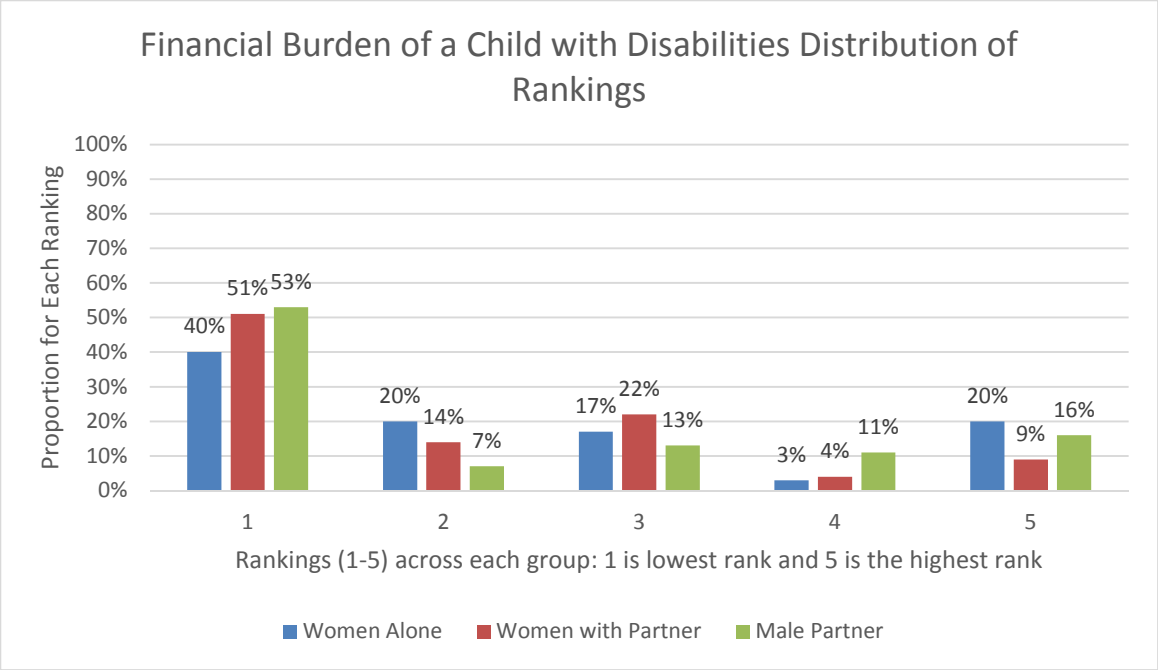

Figure 6. Proportions of each individual and their rankings (1 through 5) on the impact that a potential financial burden of a child with disabilities had on their genetic testing decision

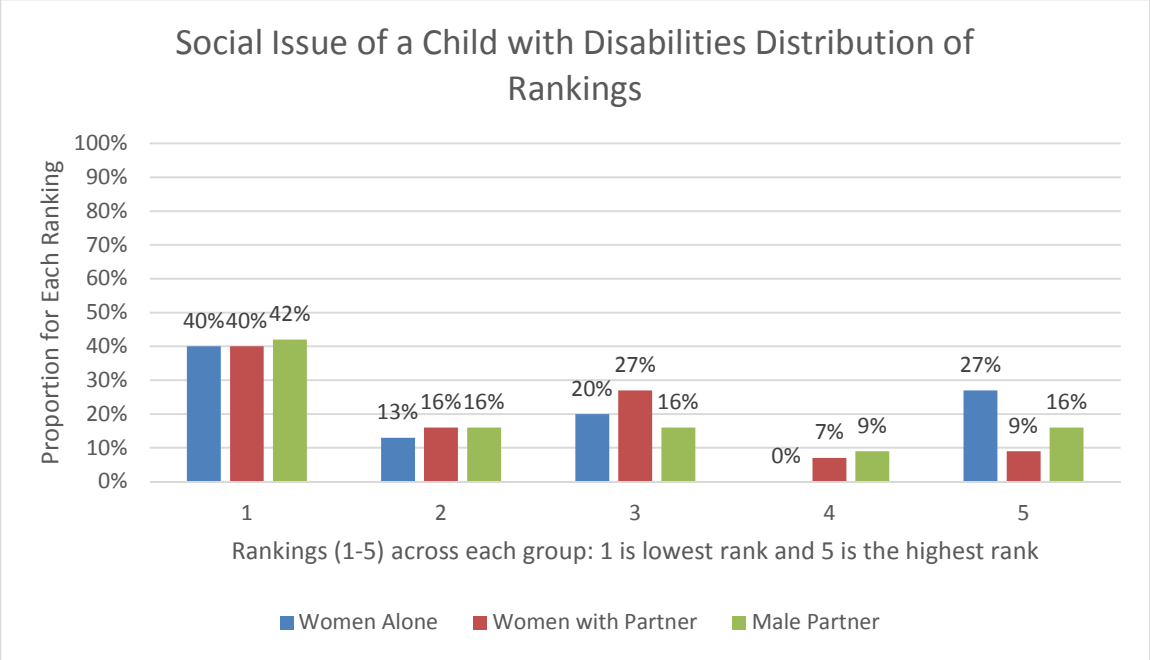

Figure 7. Proportions of each individual and their rankings (1 through 5) on the impact that a potential social issue of raising a child with disabilities had on their genetic testing decision

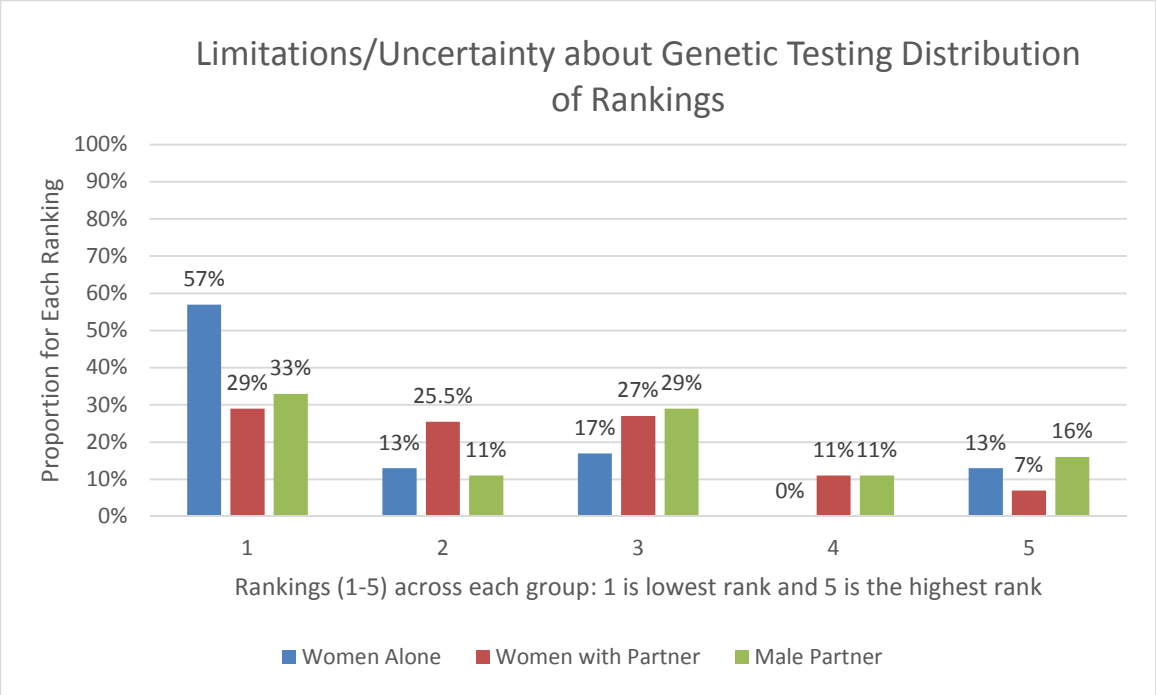

Figure 8. Proportions of each individual and their rankings (1 through 5) on the impact that the limitations or uncertainty about genetic testing had on their genetic testing decision

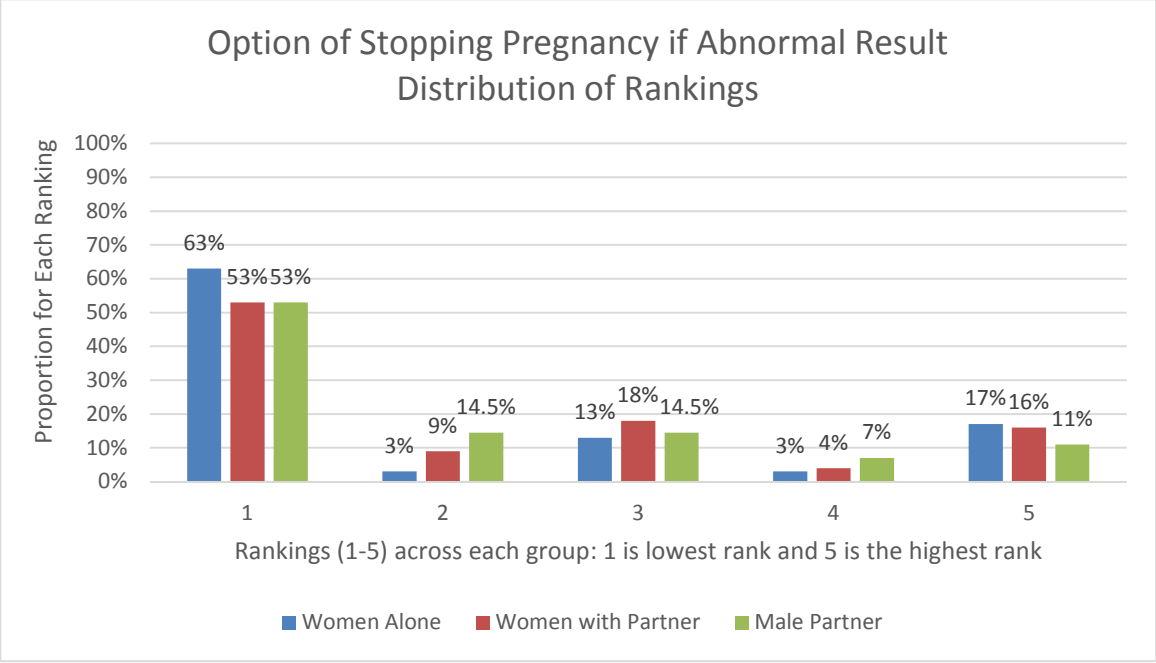

Figure 9. Proportions of each individual and their rankings (1 through 5) on the impact that the option of stopping the pregnancy if abnormal result factor had on genetic testing decision

In order to compare the ordinal rankings between the three categorical groups, a Mann-Whitney U test was utilized (Tables 15 & 16). A two-tailed hypothesis ($H_0 \neq H_a$) and significance level (α) of 0.05 were established for the analysis. Assumptions 1, 2, 3 and 4 were all met before performing the analysis. Since the 4th assumption (distributions of rankings are the same or similar shape) was met for each factor, the analysis compared the medians of the distribution. No significant differences were noted between the two groups of women. Of note, past pregnancy history (p-value = 0.052) was more commonly ranked as a contributing factor by women who came alone compared to women who came with their partner. In addition, women who came with their partner more frequently ranked limitations or uncertainty about genetic testing higher than women who came alone (p-value = 0.058).

Factors involved in testing decision	Women who came Alone (n=30)	Women who came with Partner (n=55)	P-value (2-tailed)
Pain of Invasive Test	40.38	44.43	0.444
Risk from Invasive Test	45.00	41.91	0.508
Concern about health of pregnancy/baby	46.28	41.21	0.239
Past pregnancy history	49.43	39.49	0.052
Cost of testing/insurance worries	47.60	40.49	0.151
Financial burden of a child with disabilities	46.63	41.02	0.286
Social issue/difficulty of raising a child with disabilities	45.13	41.84	0.538
Limitations or uncertainty about genetic testing	36.42	46.59	0.058
Option of stopping pregnancy if result is abnormal	40.85	44.17	0.510

Table 14. Mann Whitney U mean rank values for each factor between both groups of women

The ranked factors between both groups of women and all male partners were also compared (Table 15). Statistical significance was detected between these two groups with regard to pain from the invasive testing as women ranked this higher than their male partners (p-value = 0.010). Of note, but not of significance, male partners commonly ranked cost of testing or insurance worries (p-value = 0.135) and limitations or uncertainty about genetic testing (p-value = 0.110) as a higher contributor to their testing decision. No

significant differences were detected between the two groups with regard to any of the other ranked factors.

Factors involved in testing decision	Both groups of women (n=85)	Male Partners (n=55)	P-value
Pain of Invasive Test	76.96	60.51	**0.010**
Risk from Invasive Test	73.32	66.15	0.229
Concern about health of pregnancy/baby	71.68	68.67	0.583
Past pregnancy history	71.21	69.40	0.775
Cost of testing/insurance worries	66.74	76.32	0.135
Financial burden of a child with disabilities	70.28	70.84	0.933
Social issue/difficulty of raising a child with disabilities	70.59	70.35	0.972
Limitations or uncertainty about genetic testing	66.26	77.05	0.110
Option of stopping pregnancy if result is abnormal	70.49	70.51	0.998

Table 15. Mann Whitney ranked means between women who came with their partner and their male partners ranked factors

Women who came with their partner and their male partners were compared in a matched-pairs analysis (Table 16). A Wilcoxon Matched-Pairs Signed-Ranked test was used to determine significance. The mean (average) rank value for each factor between the two groups is listed in the table below. Statistical significance was detected within each couple with regard to pain of the invasive test (0.006), risk from the invasive test (0.041) and cost

of testing/insurance worries (0.016). Women in the couple most commonly ranked pain and risk from the invasive test (amniocentesis or CVS) higher than their male partners. The male partners commonly ranked cost of testing or insurance worries higher than their female partners.

Factors involved in testing decision	Women who came with Partner (n=55)	Male Partners (n=55)	P-value
Pain of Invasive Test	2.54	1.78	**0.006**
Risk from Invasive Test	4.27	3.84	**0.041**
Concern about health of pregnancy/baby	4.36	4.38	0.921
Past pregnancy history	2.04	2.16	0.600
Cost of testing/insurance worries	1.76	2.27	**0.016**
Financial burden of a child with disabilities	2.05	2.31	0.211
Social issue/difficulty of raising a child with disabilities	2.29	2.42	0.518
Limitations or uncertainty about genetic testing	2.42	2.67	0.314
Option of stopping pregnancy if result is abnormal	2.22	2.09	0.495

Table 16. Matched pairs analysis of women with partner and partner's responses to ranked factors

For each group, the women and the male partners were both asked to independently rank each factor that they believed their partners were most influenced by. Women who came alone ranked the factors that they believe their male partners would

have been influenced by had their partner attended the session. A Mann-Whitney U test with a two-tailed hypothesis ($H_0 \neq H_a$) and significance level (α) of 0.05 will be used to assess for statistical differences between the two groups. The mean rank values computed by the Mann-Whitney U test are listed in the tables for each nominal group. Table 17 compares the male partners' self-reported rankings and the rankings that all women reported of their male partners. Table 18 compares the women's self-reported rankings and the perceived rankings that the male partners who came to the session reported of their female partners.

No significant differences were detected between male partners' self-reported rankings for each factor (Table 17) and what women believed their male partners rankings would be. Of note, but not statistically significant, women commonly believed their male partners would rank cost of testing or insurance worries (p-value = 0.130), financial burden of a child with disabilities (p-value = 0.109) and limitations or uncertainty of genetic testing (p-value = 0.168) higher than the actual rankings that these men gave.

Factors involved in testing decision	Women's perceived ranks of their male partner (n=85)	Male partners' ranks (n=55)	P-value
Pain of Invasive Test	68.54	73.54	0.378
Risk from Invasive Test	69.11	72.65	0.578
Concern about health of pregnancy/baby	71.42	69.07	0.668
Past pregnancy history	68.88	73.00	0.506
Cost of testing/insurance worries	74.50	64.32	0.130
Financial burden of a child with disabilities	74.74	63.95	0.109
Social issue/difficulty of raising a child with disabilities	72.14	67.97	0.539
Limitations or uncertainty about genetic testing	74.21	64.76	0.168
Option of stopping pregnancy if result is abnormal	69.45	72.12	0.675

Table 17. Mann-Whitney U mean rank value of women's perceived rankings for their male partners and male partner's self-reported rankings

No significant differences were detected between women's self-reported rankings for each factor (Table 18) and what their male partners believed that the women would rank as factors affecting their genetic testing decision. Of note, but not statistically significant, male partners commonly believed their female partners would rank concern about the health of pregnancy or baby lower (p-value = 0.176) than the actual rankings the female partners gave.

Factors involved in testing decision	Male partner's perceived ranks for their female partner (n=55)	Female's actual ranks (n=85)	P-value
Pain of Invasive Test	74.93	67.94	0.275
Risk from Invasive Test	70.91	70.24	0.908
Concern about health of pregnancy/baby	65.84	73.52	0.176
Past pregnancy history	71.85	69.63	0.730
Cost of testing/insurance worries	69.47	71.16	0.784
Financial burden of a child with disabilities	68.70	71.66	0.650
Social issue/difficulty of raising a child with disabilities	72.76	69.04	0.578
Limitations or uncertainty about genetic testing	66.57	73.04	0.333
Option of stopping pregnancy if result is abnormal	70.23	70.68	0.944

Table 18. Mann-Whitney U mean rank values for each factor that affected testing decision with male partner's perceived rankings of the women and women's self-reported ranked factors

Using these same ranked factors, comparisons were made between the two groups of women to determine if there were significant differences in the ranked factors between women who elected testing and those who declined testing. A Mann-Whitney U test with a two-tailed hypothesis ($H_0 \neq H_a$) and significance level (α) of 0.05 was used to determine statistical significance between the two groups. The mean rank values from the Mann-

Whitney U test are listed in the table below. Women who elected genetic testing ranked concern about health of pregnancy or baby (p-value = 0.001) and limitations or uncertainty about genetic testing (p-value = 0.035) significantly higher than those who declined genetic testing. Of note, but not of statistical significance, women who elected genetic testing ranked social issue or difficulty of raising a child with disabilities higher (p-value = 0.060) than those who declined genetic testing.

Factors involved in testing decision	Elected Genetic Testing (n=44)	Declined Genetic Testing (n=41)	P-value
Pain of Invasive Test	40.73	45.44	0.351
Risk from Invasive Test	43.02	42.98	0.992
Concern about health of pregnancy/baby	49.50	36.02	**0.001**
Past pregnancy history	44.03	41.89	0.661
Cost of testing/insurance worries	46.85	38.87	0.091
Financial burden of a child with disabilities	44.15	41.77	0.636
Social issue/difficulty of raising a child with disabilities	47.65	38.01	0.060
Limitations or uncertainty about genetic testing	48.23	37.39	**0.035**
Option of stopping pregnancy if result is abnormal	45.76	40.04	0.235

Table 19. Factors which contributed to decision to elect or decline genetic testing for women who came alone and women who came with their partner

4. Male Partner Involvement and Decision-Making

The impact that each male partner had on the genetic testing decision was measured ranging from no influence to high influence by each respondent (Table 20). Women who came alone were asked how much of an influence their partner impacted the testing decision, even without being present. Women who came with their partner were asked to mark the level of influence that their male partner had on the genetic testing decision. Male partners were asked to mark the level of influence that they believed that they had on this testing decision. A Mann-Whitney U test with a two-tailed hypothesis ($H_0 \neq H_a$) and significance level (α) of 0.05 between the two groups of women and between the women with partner and male partners was used to assess for statistical differences. Statistical significance was detected between the women alone and women with partner groups (p-value = 0.041). Statistical significance was not detected between the women with partner and male partner groups (p-value = 0.570).

	Women Alone (n=30)		**Women with Partner (n=55)**		**Male Partner (n=55)**	
Level of influence	N	%	N	%	N	%
No influence	10	33.3%	9	16.4%	4	7.3%
Low influence	5	16.7%	12	21.8%	11	20%
Moderate influence	10	33.3%	12	21.8%	20	36.4%
High influence	5	16.7%	22	40%	20	36.4%

Table 20. Level of influence male partner had on testing decision based on woman's report and male partner's self-report

Women who came alone were asked why their partner missed the genetic counseling appointment. The "other" option included a write-in section, in which common answers included: home with kids or prison. The partners who missed the appointment were absent due to work most (80%) of the time.

	Partner's reason for missing appointment (n=30)	
Reason for missing appointment	N	%
Work	24	80%
Separated	1	3.3%
Illness/Injury	1	3.3%
Other	4	13.3%

Table 21. Reason for male partner not present at session (mother alone only)

Women who came alone were asked how helpful it would have been to have their male partner present. Women who came with their partner were asked how helpful it was to have their male partner present and their partners were asked how helpful it was for them to be present at the session (Table 22). A Mann-Whitney U test with a two-tailed hypothesis ($H_0 \neq H_a$) and significance level (α) of 0.05 was used to determine statistical significance. Statistical significance (p-value = 0.0001) was detected between the two groups of women. Women alone responded that having their partner at the session would have been helpful 30% of the time while women who came with their partner reported that having their partner present was helpful 83.6% of the time. Male partners also reported their presence was very helpful a large proportion (78.2%) of the time.

	Women Alone (n=30)		Women with Partner (n=55)		Male Partner (n=55)	
Level of utility	N	%	N	%	N	%
Not helpful	10	33.3%	0	0%	0	0%
A little helpful	7	23.3%	1	1.8%	2	3.6%
Moderately helpful	4	13.3%	8	14.5%	10	18.2%
Very helpful	9	30%	46	83.6%	43	78.2%

Table 22. Level of utility for male partner's presence at genetic counseling session

All participants were asked to select who is the primary decision-maker in pregnancy (Table 23) and who is the primary decision-maker outside of pregnancy (where to eat, live, travel, etc.) (Table 24). A Mann-Whitney U test with a two-tailed hypothesis ($H_0 \neq H_a$) and significance level (α) of 0.05 was used to detect any differences in between

the two groups of women based off a ranking system for who is the decision-maker in pregnancy. Statistical significance was detected between the two groups of women with the women alone more commonly selecting that they are the primary decision-makers in pregnancy (p-value = 0.003) and in the relationship (p-value = 0.004).

	Women Alone (n=30)		**Women with Partner (n=55)**		**Male Partner (n=55)**	
Decision-maker in pregnancy	N	%	N	%	N	%
"I do"	12	40%	8	14.5%	3	5.5%
"Equally"	18	60%	42	76.4%	44	80%
"my partner does"	0	0%	5	9.1%	8	14.5%

Table 23. Comparing who in relationship is primary decision maker in pregnancy

	Women Alone (n=30)		**Women with Partner (n=55)**		**Male Partner (n=55)**	
Decision maker outside of pregnancy	N	%	N	%	N	%
"Always me"	1	3.3%	1	1.8%	3	5.5%
"Usually me"	8	26.7%	3	5.5%	9	16.4%
"Equal"	20	66.7%	43	78.2%	38	69.1%
"Usually my partner"	0	0%	8	14.5%	5	9.1%
"Always my partner"	1	3.3%	0	0%	0	0%

Table 24. Comparing who in relationship is primary decision maker outside of pregnancy

Each participant was asked to rank the level of involvement of their male partners in past pregnancies (Table 25). The male partner respondents were asked to rank their own perceived level of involvement while the two groups of women ranked how involved they believed their male partners were. The "N/A" group refers to male partners in which there were no past pregnancies to be involved in or the question in general is not applicable to this current male partner. A Mann-Whitney U test with a two-tailed hypothesis ($H_0 \neq H_a$) and significance level (α) of 0.05 was used to determine statistical significance between the

three groups. No statistical significance was detected between the two groups of women (p-value = 0.935).

	Women Alone (n=30)		Women with Partner (n=55)		Male Partner (n=55)	
Level of involvement of male partner	N	%	N	%	N	%
Not involved	2	6.7%	3	5.5%	1	1.8%
A little involved	6	20%	7	12.7%	3	5.5%
Moderately involved	9	30%	5	9.1%	10	18.2%
Very involved	10	33.3%	26	47.3%	28	50.9%
N/A	3	10%	14	25.5%	13	23.6%

Table 25. Level of involvement in past pregnancies

All participants were asked to select their support groups (as many as they wished) in decision-making. All three groups most commonly ranked their partner as a support group with 83.3% for women alone, 90.9% of women with their partner and 92.7% of male partners ranked the partner as a support group.

	Women Alone (n=30)		Women with Partner (n=55)		Male Partner (n=55)	
Support Groups	N	%	N	%	N	%
Partner	25	83.3%	50	90.9%	51	92.7%
Friends	12	40%	22	40%	22	40%
Family	24	80%	40	72.7%	42	76.4%
Doctor/health care provider	7	23.3%	28	50.9%	26	47.2%
Church/faith	9	30%	17	30.9%	11	20%
Pets	2	6.7%	3	5.5%	4	7.3%
Social Media/TV/Internet	2	6.7%	1	1.8%	4	7.3%
Other	0	0%	2	3.6%	1	1.8%

Table 26. Reported support systems in making difficult decisions for each group

DISCUSSION

1. Background/Demographic Information

The goal of this project was to determine how influential the presence of the male partner at the genetic counseling session is to a woman's ability to make prenatal genetic testing decisions. In addition, the project aimed to elucidate the factors that impacted how male partners approach prenatal genetic testing decisions and their impact on the shared decision-making dynamic. Information gained from this study can be utilized to help prenatal genetic counselors better understand the influential factors impacting both women who come alone and couples when making prenatal genetic testing decisions. Based off of prior literature and clinical experience, we hypothesized that women who attend with their partners will be more likely to elect genetic testing, including invasive testing, when compared to women who come without their partners. Women who attend genetic counseling alone were also expected to act as the primary decision-maker in genetic testing decisions and in other decisions during the pregnancy.

A total of 140 respondents (30 women who came alone and 55 couples) completed the survey in entirety. The respondents were divided into groups based on the presence or absence of the male partner at the genetic counseling session. The three groups included: women who came alone, women who came with their partner, and the male partners in attendance. Women who came alone were significantly ($p = 0.016$) (Table 7) less likely to be married and significantly more likely to be seen at UCIMC ($p = 0.013$) (Table 8) compared to the women who came with their partners. One might expect that women who come alone to the appointment are less likely to be married. However, this does not mean that most women who come alone are single, as only 13% of these women reported as

being single. Therefore, the majority of women, regardless of whether their partners attend the session, responded as in a relationship of some kind. Prenatal genetic counselors, regardless of whether the woman has come alone or not, should not make assumptions about the relationship status of their patient, although they can expect to see women who come alone be less likely to report being married. As for the recruitment site disparity, SJMC typically sees a more affluent patient population while UCIMC has a lower socioeconomic population which may have altered the results, as this was not assessed with the survey. No other significant demographic differences were detected between women who came alone and women who came with their partner regarding the following: age, ethnicity, education level, number of prior pregnancies, number of children or referral indication.

For the male partners, no significant differences were detected between the male partners who attended the session and the male partners who could not attend with regard to age or ethnicity. Male partners who attended the genetic counseling session were significantly ($p = 0.008$) (Table 4) more likely to have a college degree compared to the male partners who did not attend. For individuals with a lower education level, it is possible that requesting time off of work may be more of a challenge, as 80% of the male partners who missed the appointment did so because of work (Table 21). The male partners who attend the genetic counseling session may do so because they are more likely to possess the financial security to request time away from work. One can expect that most male partners will miss the appointment due to their work. Asking women why their partner is not in attendance may not typically be performed during the genetic counseling

session, although if the topic does arise, prenatal genetic counselors should expect that the majority of the time, the male partner may be absent because of work.

The first hypothesis analyzed was to compare how often the two groups of women (women who came alone and women who came with their partner) elected genetic testing (Table 10). Both groups of women were asked to mark all genetic test choices elected amongst the following options: NIPS, chorionic villus sampling with chromosome analysis, amniocentesis with chromosome analysis, and "other" which included a write-in option. Women who came alone and women who came with their partner revealed no significant difference in frequency of genetic testing ordered or frequency of invasive testing ordered (Table 10 & 11). In addition, both groups of women were asked which testing they would have been comfortable making in the presence or absence of their male partner (Table 12 & 13). 33.3% of women who came alone and declined testing reported feeling comfortable with an invasive test if their male partner were present. Only 13.8% of women who came with their partner and elected testing would have still considered an invasive test without their partner present. Although no significant differences were detected between the two groups of women regarding genetic testing and invasive testing choices, women do report feeling more comfortable making an invasive testing decision with their partner present. Future studies with a larger sample size may help further clarify these findings. However, level of comfort with a testing decision does not necessarily reflect what the women would have realistically elected as there are many other factors that influence how women make genetic testing and invasive testing decisions. In addition, questions that pose hypothetical situations such as these are only exploratory but may help guide prenatal genetic counselors when approaching women and couples about genetic testing decisions. Due to

the complexity of making an invasive testing decision, hypothetically knowing that women feel more comfortable about making an invasive testing choice in their partner's presence can provide insight for prenatal genetic counselors. We recommend continuing to engage women in approaching genetic testing decisions by evaluating all relevant factors and to consider that women who come alone may wish to consult their male partners or have them present before considering an invasive testing option.

2. Factors Influencing Genetic Testing Decision

To analyze the factors involved in making prenatal genetic testing decisions, the following factors were ranked by each group of respondents: pain from invasive test, risk from invasive test, concern about health of pregnancy or baby, past pregnancy history, cost of testing or insurance worries, financial burden of a child with disabilities, social issue of raising a child with disabilities, limitations or uncertainty about genetic testing, and option of stopping the pregnancy if there is an abnormal result(Graphs 1-9). The listed factors analyzed were chosen from previous studies. [3,4,65]

When the ranked factors were compared between the two groups of women, no significant differences were detected between any factors. Although, past pregnancy history was more likely to be a highly ranked factor by women who came alone compared to women who came with their partner, regardless of whether testing was elected or declined ($p = 0.052$) (Table 14). It is unclear why past pregnancy history was ranked higher for women who came alone, although it may be due to the small sample size. It is possible that past pregnancy history may be connected to why these women have come to the appointment alone. Also, women who came with their partner were more likely to rank

limitations or uncertainty about genetic testing higher ($p = 0.058$). Women who came with their partner possibly ranked limitations or uncertainty about genetic testing higher due to interactions with their male partners. Male partners frequently ranked limitations or uncertainty about genetic testing highly with 16% ranking this factor as a 5 and only 7% of women with their partner ranking this as a 5 (Graph 8). Previous studies have also reported that male partners are concerned about limitations or uncertainty about genetic testing, and thus, may have influenced the thought process for these women who came with their partner when approaching this survey.[62,65] Aside from these two factors, both groups of women approach the other factors similarly about making genetic testing decisions. Since no statistically significant differences were detected, prenatal genetic counselors should expect all women to be influenced by similar factors. Thus, prenatal genetic counselors should continue to focus on exploring how women make decisions and what factors are most important to them on a case by case basis.

The same ranked factors were compared between all male partners who answered surveys and both groups of women (Table 15) to determine significant differences between the two genders. Women commonly ranked the pain of invasive testing significantly higher than men ($p = 0.010$), which is understandable given that the women would have to endure both the physical and emotional pain of the invasive test, while men only endure the emotional pain. However, this survey did not elucidate whether these women had previously had an invasive test or whether they were referring to emotional or physical pain of the test. To further clarify how couples evaluate genetic testing within the shared decision-making dynamic, a matched-pairs analysis was used to directly compare factors within each couple (Table 16). Again, a significant difference was detected between women

and their male partners with regard to pain from an invasive test ($p = 0.006$). In addition, women within the couple ranked the risk from invasive testing significantly higher than their male partners ($p = 0.041$). This significant difference could be due to how women perceive the risk for miscarriage. With women the experience of pregnancy is physical, and the emotional toll of a miscarriage can be more emotionally or physically challenging than for their male partners.[61,63,67]

In addition, men ranked limitations or uncertainty about genetic testing ($p = 0.110$) and cost of testing or insurance worries ($p = 0.135$) higher, but not significantly higher when compared as a group to all women. Given that genetic testing is often costly, and that screening tests often come with limitations and uncertainty, men may be more likely to avoid genetic testing unless these two factors are not of concern in their situation or if the benefits of testing outweigh these costs. Within the matched-pairs analysis, men ranked cost of testing or insurance worries significantly higher ($p = 0.016$) than their female partners. Male partners have ranked fiscal reasons for genetic testing higher than their female partners possibly due to some men feeling as if they must be the financial support system for a family.[65,66] Prenatal genetic counselors are trained in helping couples explore their options and how they make decisions. With this information, prenatal genetic counselors can expect to see women within this shared decision-making dynamic focus on the pain and risk of an invasive test while men focus on fiscal challenges or limitations of genetic testing. To help clarify these factors for couples, exploring the risk of miscarriage in several different numerical representations (such as fractions, percentages or more visual representations of the risk probability) may be helpful. Also, explaining the pain of an invasive test in a way in that women may connect and understand the pain, both emotional

and physical, can be considered by prenatal genetic counselors. Finally, opening the discussion regarding the cost or limitations of testing is always important with genetic testing in general, but may be of particular benefit for the men in the couple to hear.

In addition to respondents ranking the factors that affected how they make decisions, each respondent was asked to rank the factors that they perceived to impact their partners. Women's perceived rankings of their male partners were then compared to male partner's actual ranking (Table 17). Women commonly perceived that their male partners would rank cost of testing or insurance worries ($p = 0.130$), financial burden of a child with disabilities ($p = 0.109$) and limitations or uncertainty about genetic testing ($p = 0.168$) higher than what their male partners actually chose to rank for these factors. While none of these differences were significant, it does point to the possibility that women are highly aware of their male partner's concerns with genetic testing, but possibly overestimate the extent of that concern. Men's perceived rankings of their female partners were then compared to the women's actual rankings (Table 18). No significant or noticeable differences were detected with this analysis. Essentially, this confirms that men and women both appear to understand the factors that affect each other's genetic testing decisions.

The final factors considered were those between women who elected any genetic testing and women who declined all genetic testing. Women who elected genetic testing ranked concern about the health of pregnancy or baby ($p = 0.001$) and limitations or uncertainty about genetic testing ($p = 0.035$) significantly higher than women who declined all testing. For women who are concerned about the health of their pregnancy or baby, it would make sense that they would be interested in genetic testing to evaluate the health of

this baby using any testing available. With regard to limitations or uncertainty of genetic testing, women who have elected genetic testing would likely be more concerned by the limitations or uncertainty of the testing given that the majority of the women who elected testing chose NIPS (93%) (Table 11). Screening tests, such as NIPS, always involve a level of uncertainty for patients but provide a non-invasive approach without risks to the pregnancy.[11,32,33] Although, it is striking that women who declined testing ranked this significantly lower. This may be due to a disinterest in all genetic testing that prevented them from ever considering the limitations of these screening tests as a crucial factor.

3. Male Partner Involvement and Decision-Making

Understanding how the male partner impacts genetic testing decisions extends further beyond the actual genetic testing elected as genetic testing decisions involve a vast array of factors. Another question that this study aimed to answer was the level of comfort and decision-making dynamics between women who come alone and couples. We hypothesized that women who come alone are confident making decisions on their own while women who come with their partner will seek out guidance from their male partners. Survey respondents were asked how much influence their partner had on their genetic testing decision. Women who came alone reported that their male partners had no influence 33.3% of the time compared to only 16.4% of women who came with their partner (Table 20). 40% of women who came with their partner reported that the male partner had a high influence on the testing decision compared to only 16.7% of women who came alone. When the male partners who attended the session were asked the same question, they responded that they had no influence 7.3% of the time and high influence

36.4% of the time. Based on the scaled responses between "no influence" and "high influence," women who came with their partner responded that their male partners had a significant impact on the genetic testing decision ($p = 0.041$). While no significant difference was detected in proportion of genetic testing ordered, these responses do point toward the male partner presence impacting genetic testing decision-making.

In addition to measuring the level of impact the male partners had on genetic testing, the level of utility to the genetic counseling session was measured. Women who came alone responded that their male partners would not have been helpful to the session 33.3% of the time and would have been very helpful 30% of the time (Table 22). In contrast, women who came with their partner reported the male partner was not helpful 0% of the time and very helpful 83.6% of the time. These responses also show a significant difference between women who came alone and women who came with their partner about how helpful the male partners were or would have been to the genetic counseling session ($p = 0.0001$). When male partners were asked how helpful they thought it was to be at the session, 0% responded "not helpful" and 78.2% responded "very helpful". This only further clarifies the influence that male partners had on the genetic counseling appointment. Male partners frequently replied how helpful their presence was to the appointment which also supports their presence at the appointment. Since women who came alone frequently reported that their male partners' presence would not have been helpful, it possibly points to the idea that women who came alone either do not have an involved male partner, do not want their male partners present, or feel confident making any decisions on their own.

To further elucidate decision-making dynamics for these two groups of women, questions were asked to explore decision-making within the pregnancy and within the relationship. First, respondents were asked who the primary decision-maker in pregnancy is and then were asked who the primary decision-maker in the relationship is for decisions such as: where to eat, where to live, traveling decisions, etc. Women who came alone responded as the primary decision-maker 40% of the time and women who came with their partner responded as the primary decision-maker 14.5% of the time (Table 23). None (0%) of the women who came alone answered that their partner is the primary-decision maker while 9.1% of women who came with their partner responded this way. Women who came alone were significantly more likely to be the primary decision-maker in the pregnancy ($p = 0.003$). In addition, women who came alone were found to be significantly more likely to be the primary decision-maker in relationship decisions ($p = 0.004$) (Table 24). Both analyses continue to support the stated hypothesis that women who come alone are confident making decisions on their own. In addition, women who came with their partner reported that pregnancy and relationship decision-making is equal 76.4% and 78.2% of the time, respectively. This data further supports the hypothesis that women who bring their partners are more likely to engage in a shared decision-making dynamic.

In conclusion, prenatal genetic counselors can feel confident that women who come alone are able to make genetic testing decisions without their male partners' support. Also, women who come with their partner often have their partner at the genetic counseling appointment because they wish to make decisions together. For these reasons, it is important to examine the concerns of the male partner when present as they will have an impact on the genetic testing decision that is made. As stated earlier, both the man and

woman within the couple are influenced by distinct factors, so prenatal genetic counselors should work to focus on risk and pain from invasive testing for women and the cost of testing or insurance worries for the male partners. While no significant difference was detected in the frequency and proportion of genetic tests elected between women who come alone and women who come with their partner, prenatal genetic counselors should explore these factors to better understand how patients are influenced.

4. Conclusion & Future Considerations

Furthering our understanding of how women and couples make prenatal genetic testing decisions will help the field progress in our ability to counsel patients as well as organize genetic testing options for patients. One of the hypotheses that could not be successfully analyzed, given the time constraints and sample size, was the frequency of invasive testing choices made by women who came alone and women who came with their partners. To better evaluate this hypothesis, a more robust study is necessary. Future studies may also consider analyzing how women who came alone and couples differ in carrier screening testing choices. One may expect that given the nature of carrier testing for autosomal recessive diseases, in which both parents would have to be carriers to have an affected child, carrier testing choices may differ in the presence of the male partner.

The design of this study utilized a collection of factors that are known to have an impact on how women and couples approach prenatal genetic testing decisions. However, most of these factors were generated from surveys in which the primary respondents were women. A few studies have examined and explored the role men play in prenatal screening and genetic testing, yet these studies were limited in proposing factors that influenced

genetic testing decision-making.[62,64,65] Thus, it may be informative to conduct a more exploratory study into factors not considered by this or past research to impact the decision making of men. By doing so, the role of the male partner and their influence on genetic testing decisions within a couple's shared decision-making dynamic can be best defined. Another option to further the analysis of the male partner influence would be to survey the male partners who were unable to attend. Research of this nature may help us determine the underlying reasons for why male partners do not attend genetic counseling. Surveying these men could also act as an opportunity to evaluate the factors that they may consider when making genetic testing decisions.

Survey analysis can only elucidate the surface response of patients who present to genetic counseling, so more in-depth analysis of decision-making is warranted. We propose evaluating the level of confidence, anxiety, certainty, or apathy of women and couples for genetic testing decision-making. Further analysis will help clarify decision-making roles in the pregnancy and allow prenatal genetic counselors to better understand the needs of their patients. In addition, exploring how women and couples prefer to be approached regarding genetic testing decision-making will be useful in guiding this field further.

To summarize, this study identified that women who come alone feel confident making genetic testing decisions independently of their male partners while women who come with their partner prefer a shared decision-making dynamic. Within this shared decision-making dynamic, women are more concerned about risk and pain of invasive testing while men are more concerned about the cost of testing or insurance coverage for testing. In addition, both groups of women elected genetic testing and invasive testing at the same rate. Gathering this information serves as an important tool for prenatal genetic

counselors to adapt to the needs of the patient and individualize their care. In the world of precision medicine, it is vital to personalize all medical services, especially genetic counseling, to the needs of the men and women who present to genetic counseling. With the ever-increasing number of genetic testing choices, prenatal genetic counselors will continue to be the bridge for patients in helping them make a truly informed decision that caters to their beliefs and concerns with the pregnancy.

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APPENDIX A

UCI University of California, Irvine

CONFIRMATION OF EXEMPT RESEARCH REGISTRATION

November 21, 2017

DILLON NORRIS VAN DEN BERG
PEDIATRICS - GENETICS

RE: HS# 2017-3913 *Affect of Male Partner's Involvement in a Woman's Prenatal Decision Making Process*

The human subjects research project referenced above has been administratively registered with the UC Irvine Institutional Review Board (UCI IRB) as Exempt from Federal regulations in accordance with 45 CFR 46.101. This exemption is limited to the described activities in the registered UCI IRB Protocol Narrative and extends to the performance of such activities at the sites identified in your UCI IRB Protocol Application. Informed consent from subjects must be obtained unless otherwise indicated below. UCI IRB conditions for the conduct of this research are included on the attached sheet.

Information provided to prospective subjects to obtain their informed consent should, at a minimum, consists of the following information: the subject is being asked to participate in research, what his/her participation will involve, all foreseeable risks and benefits, the extent to which privacy and confidentiality will be protected, that participation in research is voluntary and the subject may refuse to participate or withdraw at any time without prejudice.

Questions concerning registration of this study may be directed to the UC Irvine Office of Research, 141 Innovation Drive, Suite 250, Irvine CA 92697-7600; 949-824-0665 (biomedical committee) or 949-824-6662 (social-behavioral committee).

Level of Review: Exempt Review, Category 2

Cristobal Barrios, MD
Vice Chair, Institutional Review Board
Registration valid from 11/21/2017 to 11/20/2022
UCI (FWA) 00004071, Approved: January 31, 2003

Determinations as Conditions of Exemption:
Informed Consent Requirements:

1. Signed Informed Consent Not Required
    a. Study Information Sheet Required
2. Use of Translated Language Consent[1]

APPENDIX B

University of California, Irvine
Study Information Sheet

Effect of Male Partner's Involvement in a Woman's Prenatal Decision-Making Process

Lead Researcher
Dillon van den Berg, Genetic Counseling Intern
Department of Pediatrics & Genetics
(714) 456-5837 or dvandenb@uci.edu

Faculty Sponsor
Kathyrn Steinhaus-French, Clinical Professor & Genetic Counselor
Department of Pediatrics & Genetics
(714) 456-6873 or kasteinh@uci.edu

- We are asking you to take part in a study conducted by researchers at the University of California, Irvine. Participating in this study is optional.
- If you choose to be in the study, you will complete a survey. Questions will be asked about how you and your partner make decisions with regards to genetic testing. This survey will help us learn more about the factors that contribute to how a couple makes testing decisions and further understand the dynamics of how a relationship impacts these testing decisions. The survey will take about 5-10 minutes to complete.
- You can skip questions that you do not want to answer or stop the survey at any time. The survey is anonymous, and no one will be able to link your answers back to you. Please do not include your name or other information that could be used to identify you in the survey responses. We will keep your answers confidential and will not share your personal information with anyone outside the research team.
- If you have any comments, concerns, or questions regarding the conduct of this research please contact the researchers listed at the top of this form.
- If you have questions or concerns about your rights as a research participant, you can contact the UCI Institutional Review Board by phone, (949) 824-6662, by e-mail at IRB@research.uci.edu or at 141 Innovation, Suite 250, Irvine, CA 92697.

  What is an IRB? An Institutional Review Board (IRB) is a committee made up of scientists and non-scientists. The IRB's role is to protect the rights and welfare of human subjects involved in research. The IRB also assures that the research complies with applicable regulations, laws, and institutional policies.

APPENDIX C

University of California, Irvine
Hoja de información del estudio

Efecto de la participación de la pareja masculina en el proceso de la toma de decisión prenatal de la mujer

Jefe de investigación
Dillon van den Berg, interno de asesoramiento genético
Departamento de Pediatría y Genética
(714) 456-5837 o dvandenb@uci.edu

Patrocinador de la facultad
Kathyrn Steinhaus-French, profesora clínica y asesora genética
Departamento de Pediatría y Genética
(714) 456-6873 o kasteinh@uci.edu

- Lo invitamos a que participe en un estudio llevado a cabo por investigadores de la Universidad de California (University of California) en Irvine. La participación en este estudio es opcional.
- Si elige formar parte del estudio, deberá completar una encuesta. Las preguntas serán acerca de cómo usted y su pareja toman decisiones respecto al examen genético. Esta encuesta nos ayudará a conocer más acerca de los factores que influyen en la toma de decisiones que realizan las parejas y nos permitirá entender más sobre las dinámicas de cómo una relación impacta en estas decisiones de exámenes. Completar esta encuesta le llevará alrededor de 5 a 10 minutos.
- Puede omitir cualquier pregunta que no desee contestar o abandonar la encuesta en cualquier momento. La encuesta es anónima, y nadie podrá vincular las respuestas con la persona que las proporcionó. No incluya su nombre ni otro tipo de información que podría usarse para identificarlo en las respuestas de la encuesta. Mantendremos sus respuestas de manera confidencial y no las compartiremos con nadie que no forme parte del equipo de investigación.
- Si tiene algún comentario, inquietud o pregunta respecto al procesamiento de esta investigación, contacte a los investigadores mencionados al comienzo de esta hoja.
- Si tiene alguna pregunta o inquietud acerca de sus derechos como participante de una investigación, puede contactar al Comité de Revisión Institucional de la UCI (University of California, Irvine) llamando al (949) 824-6662, escribiendo al correo electrónico IRB@research.uci.edu o dirigiéndose a 141 Innovation, Suite 250, Irvine, CA 92697.

  ¿Qué es un IRB? Un Comité de Revisión Institucional (Institutional Review Board, IRB) es un comité compuesto por científicos y no científicos. La función del IRB es proteger los derechos y el bienestar de los sujetos humanos que participan en una investigación. El IRB también asegura que la investigación cumpla con las regulaciones, leyes y políticas institucionales correspondientes.

APPENDIX D

Recruitment Script

Hello, my name is (name of researcher) and I am a (position of researcher) at University of California, Irvine in the Genetics Division. I am conducting research on how pregnant women and their partners make decisions regarding prenatal genetic testing.

Participation in this research includes taking a survey about what factors contributed to your testing decision, which will take approximately 5-10 minutes. Participation is voluntary and requires completion of all the questions in order to be accepted. If you choose to participate, all questions should be answered without input from your partner.

Any information provided on this questionnaire will remain confidential. Only the researchers will have access to the questionnaires and the information on it. No identifying information will be included on the survey and any personal information will not be revealed. The results of this research may be published in a scientific journal and/or presented at a professional meeting.

Once completed, please drop the survey into the lock box in the waiting room. The lockbox will keep all of your answers private and confidential and will be emptied regularly by only the research personnel.

If you have any questions about research, the lead researcher, Dillon van den Berg, can be reached at *(714) 456-5837* or *dvandenb@uci.edu.*"

APPENDIX E

Texto de reclutamiento

Hola, mi nombre es (nombre del investigador) y soy (posición del investigador) de la Universidad de California, Irvine en la División de Genética. Estoy realizando una investigación acerca de cómo las mujeres embarazadas y sus parejas toman decisiones con respecto al examen genético prenatal.

La participación en esta investigación implica realizar una encuesta, que le tomará alrededor de 5 o 10 minutos, sobre los factores que contribuyeron en su decisión respecto al examen. La participación es voluntaria y requiere que se completen todas las preguntas para ser aceptada. Si elige participar, deberá contestar todas las preguntas sin ninguna influencia de su pareja.

Toda la información proporcionada en el cuestionario será confidencial. Solo los investigadores podrán acceder a los cuestionarios y a la información que estos contengan. En la encuesta, no se incluirá información identificativa ni se revelará información personal. Los resultados de esta investigación podrán ser publicados en una revista científica o presentados en un encuentro profesional.

Una vez completa, deposite la encuesta en la caja de seguridad, en la sala de espera. La caja de seguridad mantendrá las respuestas de manera privada y confidencial, y solo el personal de investigación la vaciará regularmente.

Si tiene alguna duda sobre esta investigación, puede comunicarse con el jefe de investigación, Dillon van den Berg, llamando al *(714) 456-5837* o por correo electrónico: *dvandenb@uci.edu*".

APPENDIX F

St. Joseph Health

CERTIFICATE OF HRPP DETERMINATION

February 1, 2018

SJH Reference # 18-011
Protocol Title: Effect of Male Partner's Involvement in a Woman's Prenatal Decision Making Process

Dear Ms. Cindy Saunders:

This is to advise you that the above referenced research project has been presented to the St. Joseph Health System Human Research Protection Program (HRPP) Office for review, and the following action was taken with the explanation provided below:

Study Status: Exempt from IRB Review: 02/01/2018
Description: The SJH HRPP Office reviewed the above-referenced submission and determined that the study qualifies for Exemption from 45 CFR 46 regulations governing human subjects research in accordance with 45 CFR 46.101(b) under Category 2: Research involving the use of survey procedures, unless: (i) information obtained is recorded in such a manner that human subjects can be identified, directly or through identifiers linked to the subjects; and (ii) any disclosure of the human subjects' responses outside the research could reasonably place the subjects at risk of criminal or civil liability or be damaging to the subjects' financial standing, employability, or reputation.

Co-investigators approved: Dillon Van Den Berg; Kathryn Steinhaus-French

The following documents were reviewed:
Scientific Attestation dated 10Jan2018 - Mary Wickman
Application for IRB Exemption (signed 1-31-2018)
Protocol Version dated 11Jan2018
Questionnaire for Mother with Partner Present (English)
Questionnaire for Mother with Partner Present (Spanish)
Questionnaire for Mother without Partner (English)
Questionnaire for Mother without Partner (Spanish)
Questionnaire for Partner (English)
Questionnaire for Partner (Spanish)
Certificate of Accuracy dated 10Jan2018 and confirmation

Please note:
Although this study is exempt from Human Subjects Regulations found at 45 CFR 46, this project must be conducted in accordance with the Ethical Principles outlined in the Belmont Report.

If the study design or procedures change, please submit the changes to the HRPP Office at HRPP@stjoe.org. Please be aware that significant study changes may nullify the exemption and require IRB review and approval. **Please inform the HRPP Office via email or letter when you have completed your study.**

St. Joseph Health, Center for Clinical Research
3345 Michelson Drive, Suite 100 Irvine, CA 92612 (949) 381-4907
FWA00010992; IORG0007710; IRB00009257 (SJH IRB #1); IRB00009288 (SJH IRB #2)
This approval verifies the IRB operates in accordance with applicable ICH, federal, state, local and institutional regulations, and with all GCP guidelines governing institutional IRB operation.

APPENDIX G

St. Jude Medical Center
Study Information Sheet
Effect of Male Partner's Involvement in a Woman's Prenatal Decision-Making Process

Lead Researcher
Cindy Saunders, RN
cindy.saunders@stjoe.org

Co-Researcher
Dillon van den Berg, Genetic Counseling Intern
Department of Prenatal Diagnostics & Genetics
(714) 456-5837 or dvandenb@uci.edu

- We are asking you to take part in a study conducted by researchers at St. Jude Medical Center. Participating in this study is optional.
- If you choose to be in the study, you will complete a survey. Questions will be asked about how you and your partner make decisions with regards to genetic testing. This survey will help us learn more about the factors that contribute to how a couple makes testing decisions and further understand the dynamics of how a relationship impacts these testing decisions. The survey will take about 10-15 minutes to complete.
- You can skip questions that you do not want to answer or stop the survey at any time. The survey is anonymous, and no one will be able to link your answers back to you. Please do not include your name or other information that could be used to identify you in the survey responses. We will keep your answers confidential and will not share your personal information with anyone outside the research team.
- If you have any comments, concerns, or questions regarding the conduct of this research please contact the researchers listed at the top of this form.
- If you have any questions about your rights while participating in this study, or if you have any concerns regarding the conduct of this study, you may contact the St. Joseph Health Human Research Protection Program (HRPP) Office at 949-381-4907, by mail at 3345 Michelson Drive, Suite 100, Irvine, CA 92612, by email at HRPP@stjoe.org, or via the Internet at www.stjoe.org/Research.

  What is an IRB? An Institutional Review Board (IRB) is a committee made up of scientists and non-scientists. The IRB's role is to protect the rights and welfare of human subjects involved in research. The IRB also assures that the research complies with applicable regulations, laws, and institutional policies.

APPENDIX H

University of California, Irvine
Hoja de información del estudio
Efecto de la participación de la pareja masculina en el proceso de la toma de decisión prenatal de la mujer

Jefe de investigación
Cindy Saunders, RN
cindy.saunders@stjoe.org

Patrocinador de la facultad
Dillon van den Berg, interno de asesoramiento genético
Departamento de Pediatría y Genética
(714) 456-5837 o dvandenb@uci.edu

- Lo invitamos a que participe en un estudio llevado a cabo por investigadores de la Centro Médico de St. Jude. La participación en este estudio es opcional.
- Si elige formar parte del estudio, deberá completar una encuesta. Las preguntas serán acerca de cómo usted y su pareja toman decisiones respecto al examen genético. Esta encuesta nos ayudará a conocer más acerca de los factores que influyen en la toma de decisiones que realizan las parejas y nos permitirá entender más sobre las dinámicas de cómo una relación impacta en estas decisiones de exámenes. Completar esta encuesta le llevará alrededor de 5 a 10 minutos.
- Puede omitir cualquier pregunta que no desee contestar o abandonar la encuesta en cualquier momento. La encuesta es anónima, y nadie podrá vincular las respuestas con la persona que las proporcionó. No incluya su nombre ni otro tipo de información que podría usarse para identificarlo en las respuestas de la encuesta. Mantendremos sus respuestas de manera confidencial y no las compartiremos con nadie que no forme parte del equipo de investigación.
- Si tiene algún comentario, inquietud o pregunta respecto al procesamiento de esta investigación, contacte a los investigadores mencionados al comienzo de esta hoja.
- Si tiene alguna pregunta o inquietud acerca de sus derechos como participante de una investigación, puede contactar al Programa de protección de la investigación humana de St. Joseph Health (HRPP) Oficina a 949-381-4907, por correo a 3345 Michelson Drive, Suite 100, Irvine, CA 92612, por correo electronico a HRPP@stjoe.org, o por Internet a www.stjoe.org/Research.

¿Qué es un IRB? Un Comité de Revisión Institucional (Institutional Review Board, IRB) es un comité compuesto por científicos y no científicos. La función del IRB es proteger

los derechos y el bienestar de los sujetos humanos que participan en una investigación. El IRB también asegura que la investigación cumpla con las regulaciones, leyes y políticas institucionales correspondientes.

APPENDIX I

Recruitment Script

" Hello, my name is [name of researcher] and I am a [position of researcher] at St. Jude Medical Center in the Prenatal Diagnostics division. I am conducting research on how pregnant women and their partners make decisions regarding prenatal genetic testing.

Participation in this research includes taking a survey about what factors contributed to your testing decision, which will take approximately 10-15 minutes. Participation is voluntary and requires completion of all the questions in order to be accepted. If you choose to participate, all questions should be answered without input from your partner.

Any information provided on this questionnaire will remain confidential. Only the researchers will have access to the questionnaires and the information on it. No identifying information will be included on the survey and any personal information will not be revealed. The results of this research may be published in a scientific journal and/or presented at a professional meeting.

Once completed, please drop the survey into the lock box in the waiting room. The lockbox will keep all of your answers private and confidential and will be emptied regularly by only the research personnel.

If you have any questions about research, the lead researcher, Dillon van den Berg, can be reached at *(714) 456-5837* or *dvandenb@uci.edu.*"

APPENDIX J

Texto de reclutamiento

"Hola, mi nombre es [nombre del investigador] y soy [posición del investigador] del Centro Médico de St. Jude en la División de Diagnóstico Prenatal. Estoy realizando una investigación acerca de cómo las mujeres embarazadas y sus parejas toman decisiones con respecto al examen genético prenatal.

La participación en esta investigación implica realizar una encuesta, que le tomará alrededor de 5 o 10 minutos, sobre los factores que contribuyeron en su decisión respecto al examen. La participación es voluntaria y requiere que se completen todas las preguntas para ser aceptada. Si elige participar, deberá contestar todas las preguntas sin ninguna influencia de su pareja.

Toda la información proporcionada en el cuestionario será confidencial. Solo los investigadores podrán acceder a los cuestionarios y a la información que estos contengan. En la encuesta, no se incluirá información identificativa ni se revelará información personal. Los resultados de esta investigación podrán ser publicados en una revista científica o presentados en un encuentro profesional.

Una vez completa, deposite la encuesta en la caja de seguridad, en la sala de espera. La caja de seguridad mantendrá las respuestas de manera privada y confidencial, y solo el personal de investigación la vaciará regularmente.

Si tiene alguna duda sobre esta investigación, puede comunicarse con el jefe de investigación, Dillon van den Berg, llamando al *(714) 456-5837* o por correo electrónico: *dvandenb@uci.edu*".

APPENDIX K

CGS CARMAZZI GLOBAL SOLUTIONS

CERTIFICATE OF ACCURACY

UNITED STATES OF AMERICA)
STATE OF CALIFORNIA)
COUNTY OF SACRAMENTO)

The undersigned, Dalileah Larios, Project Coordinator for Carmazzi Global Solutions hereby states, to the best of his knowledge and belief, that the foregoing is an accurate translation of Spanish consisting of 3 pages, and this is the last of the attached.

Final Insert IPB U.S. - Reg U Loans - 01.18_SP

Dalileah Larios
PROJECT COORDINATOR
CARMAZZI GLOBAL SOLUTIONS

BEFORE ME, A NOTARY PUBLIC IN AND FOR THE STATE OF CALIFORNIA AT LARGE PERSONALLY APPEARS DALILEAH LARIOS, WHO AFTER BEING DULY SWORN, DEPOSES AND SAYS THAT THIS IS A TRUE AND CORRECT TRANSLATION OF THE ATTACHED DOCUMENT.

SWORN TO AND SUBSCRIBED THIS 20 DAY OF Jan., 2018.

SIERRA DAWN TRUJILLO
COMM. # 2204886
NOTARY PUBLIC - CALIFORNIA
SACRAMENTO COUNTY
COMM. EXPIRES JULY 14, 2021

NOTARY PUBLIC STATE OF CALIFORNIA
AT LARGE

MY COMMISSION EXPIRES: 07/14/2021

The utmost care has been taken to ensure the accuracy of the translation. Neither Carmazzi Global Solutions nor the translator shall be liable for any damages due to negligence or error in typing or translating.

T: 1 888 452 6543 • F: 866-648-3431
info@carmazzi.com • www.carmazzi.com
1026 Florin RD # 384 Sacramento, Ca 95831

APPENDIX L

Questionnaire for Mother without Partner

Purpose: In this study, we hope to find out more about how you feel about genetic testing. This study will explore how people make choices about genetic testing during pregnancy. Participating in this study is voluntary and requires you to answer all of the questions below.

If any questions should arise, feel free to contact the researcher using the contact information below:
Dillon van den Berg, Lead Researcher
University of California, Irvine
(714) 456-5837 or dvandenb@uci.edu

Confidentiality: Any information provided on this questionnaire will remain confidential. Only the researchers will have access to the questionnaires and the information on it. No identifying information will be included on the survey and any personal information will not be revealed. The results of this research may be published in a scientific journal and/or presented at a professional meeting.

Please mark each question with a check in the appropriate box or fill in the blank if prompted.

1. Your Age Group:

☐ Under 25 ☐ 25-35 ☐ 36-45

☐ 46-55 ☐ 55+

2. Your Partner's Age Group:

☐ Under 25 ☐ 25-35 ☐ 36-45

☐ 46-55 ☐ 55+

3. Which ethnic group describes you the best? (Please only check one)

☐ African American/Black ☐ White/Caucasian ☐ Asian/Pacific Islander

☐ American Indian/Alaskan Native ☐ Hispanic/Latino ☐ Other: (please specify)_____

4. In which ethnic group does your partner associate with the most? (Please only check one)

☐ African American/Black ☐ White/Caucasian ☐ Asian/Pacific Islander

☐ American Indian/Alaskan Native ☐ Hispanic/Latino ☐ Other: (please specify)_____

5. What is the highest level of education that you have achieved?

☐ Less than a high school degree ☐ High school degree ☐ Some college

☐ College degree ☐ Postgraduate degree or higher

6. What is the highest level of education that your partner has achieved?

☐ Less than a high school degree ☐ High school degree ☐ Some college

☐ College degree ☐ Postgraduate degree or higher

7. What is your current relationship/marital status?

☐ Single ☐ Widowed ☐ Committed relationship

☐ Married ☐ Divorced ☐ Separated

8. How many times have you been pregnant previously?

☐ 0 ☐ 1 ☐ 2 ☐ 3 or more

9. How many children do you currently have?

☐ 0 ☐ 1 ☐ 2 ☐ 3 or more

10. For what reason were you seen by a Genetic Counselor today?

☐ Age (35+) ☐ Positive blood test/screen ☐ Other: _____

11. Did you decide to have genetic testing after today's session?

☐ Yes ☐ No

11a. If **NO** to the above question, **SKIP** to Question #12.

11b. If **YES** to above, which testing did you elect to have (check all that apply)?

☐ CVS with Karyotype (chromosome analysis)

☐ Amniocentesis with Karyotype (chromosome analysis)

☐ CVS with Microarray

☐ Amniocentesis with Microarray

☐ Noninvasive prenatal screening (NIPT)

☐ Other: _____

11c. If **you decide to have the test:** For each of the following factors, circle a number between 1 and 5 (1=least important & 5=very important) for how important each factor was in your testing decision. Also circle a number for how important you believe each factor was for your partner.

Circle only one number for both you and your partner for each statement:	**You** **1=least important**	**Your Partner** **5=most important**
Pain from the test (CVS or Amniocentesis):	1 2 3 4 5	1 2 3 4 5
Risk from the test (CVS or Amniocentesis):	1 2 3 4 5	1 2 3 4 5
Concern about health of pregnancy/baby:	1 2 3 4 5	1 2 3 4 5
Past pregnancy history:	1 2 3 4 5	1 2 3 4 5
Cost of testing or insurance worries:	1 2 3 4 5	1 2 3 4 5
Financial burden of a child with disabilities:	1 2 3 4 5	1 2 3 4 5
Social issue/difficulty of raising a child with disabilities:	1 2 3 4 5	1 2 3 4 5
Limitations or uncertainty about genetic testing:	1 2 3 4 5	1 2 3 4 5
Option of stopping pregnancy if result is abnormal:	1 2 3 4 5	1 2 3 4 5

11d. How much of an influence did your partner's thoughts, concerns or beliefs from the above statements have on your decision to pursue testing?

☐ No influence ☐ Low influence ☐ Moderate Influence ☐ High influence

12. If you decided NOT to have a test and answered **"NO"** to question #11: For each of the following factors, circle a number between 1 and 5 (1=least important & 5=most important) for how important each factor was in your decision **NOT** to have genetic testing. Also circle a number for how important you believe each factor was for your partner.

Circle only one number for both you and your partner for each statement:	**You** **1=least important**	**Your Partner** **5=most important**
Pain from the test (CVS or Amniocentesis):	1 2 3 4 5	1 2 3 4 5
Risk from the test (CVS or Amniocentesis):	1 2 3 4 5	1 2 3 4 5
Concern about health of pregnancy/baby:	1 2 3 4 5	1 2 3 4 5
Past pregnancy history:	1 2 3 4 5	1 2 3 4 5
Cost of testing or insurance worries:	1 2 3 4 5	1 2 3 4 5
Financial burden of a child with disabilities:	1 2 3 4 5	1 2 3 4 5
Social issue/difficulty of raising a child with disabilities:	1 2 3 4 5	1 2 3 4 5
Limitations or uncertainty about genetic testing:	1 2 3 4 5	1 2 3 4 5
Option of stopping pregnancy if result is abnormal:	1 2 3 4 5	1 2 3 4 5

12a. If you answered **"NO"** to question #11: How much of an influence did your thoughts, concerns or beliefs from the above statements have on your decision to **NOT** pursue testing?

☐ No influence ☐ Low influence ☐ Moderate Influence ☐ High influence

12b. Would you have considered genetic testing, if your partner **was** present at the genetic counseling session with you? If yes, **select all** tests that you would have felt comfortable making with your partner present:

☐ CVS with Karyotype (chromosome analysis)
☐ Amniocentesis with Karyotype (chromosome analysis)
☐ CVS with Microarray
☐ Amniocentesis with Microarray
☐ Noninvasive prenatal screening (NIPT)
☐ Other: _____
☐ No Testing

13. Why was your partner absent from the session?

☐ Work
☐ Sick/Illness/Injury
☐ Separated/Not involved
☐ Other: _____

14. Do you think it would have been helpful to have your partner in the session?

☐ Not helpful ☐ A little helpful ☐ Moderately helpful ☐ Very helpful

15. Does either you or your partner usually take the lead in decisions during this pregnancy?

☐ My partner does ☐ I take the lead ☐ We share decisions equally

16. Was your partner heavily involved in decisions with past pregnancies, such as: where you went for pregnancy care, type of delivery desired, medication taken, or decisions made when complications have arisen, etc.?

☐ Not involved ☐ A little involved ☐ Moderately involved ☐ Very involved ☐ N/A

17. Who in the relationship tends to be the primary decision maker regarding: where to eat, where to live, how to spend free time together, etc.?

☐ Always Me ☐ Usually Me ☐ Equal ☐ Usually my partner ☐ Always my partner

18. Whom do you consider to be part of your support network in making difficult decisions? (select all that apply)

☐ Partner ☐ Family

☐ Friends ☐ Doctor, nurse or other care provider

☐ Church/Faith ☐ Pets

☐ Social Media/TV/Internet ☐ Other: _____

APPENDIX M

Cuestionario para la Madre Sin Pareja

Objetivo: En este estudio, deseamos saber más acerca de cómo se siente con respecto a las pruebas genéticas. Exploraremos la forma en que las personas toman decisiones con respecto a las pruebas genéticas durante el embarazo. La participación en este estudio es voluntaria y requiere que responda todas las preguntas que se encuentran a continuación.

Si surgen preguntas, no dude en comunicarse con el investigador, utilizando la siguiente información de contacto:
Dillon van den Berg, investigador principal
University of California, Irvine
(714) 456-5837 o dvandenb@uci.edu

Confidencialidad: Toda la información proporcionada en este cuestionario será confidencial. Solo los investigadores tendrán acceso a los cuestionarios y a la información contenida en ellos. No se incluirá información de identificación en la encuesta, ni se revelará información personal. Es posible que los resultados de esta investigación se publiquen en una revista científica o se presenten en una reunión profesional.

Marque la casilla correspondiente a cada pregunta o complete el espacio cuando se solicita.

1. Su grupo etario:

☐ Menor de 25 ☐ 25-35 ☐ 36-45

☐ 46-55 ☐ Mayor de 55

2. Grupo etario de su pareja:

☐ Menor de 25 ☐ 25-35 ☐ 36-45

☐ 46-55 ☐ Mayor de 55

3. ¿Con qué grupo étnico se identifica más? (Marque solo una opción)

☐ Afroamericano/negro ☐ Blanco/caucásico ☐ Asiático/isleño del Pacífico

☐ Amerindio/nativo de Alaska ☐ Hispano/latino ☐ Otro (especifique): _____

4. ¿Con qué grupo étnico se identifica más su pareja? (Marque solo una opción)

☐ Afroamericano/negro ☐ Blanco/caucásico ☐ Asiático/isleño del Pacífico

☐ Amerindio/nativo de Alaska ☐ Hispano/latino ☐ Otro (especifique): _____

5. ¿Cuál es el nivel de educación superior que completó?

☐ Inferior al diploma de la escuela secundaria ☐ Diploma de la escuela secundaria ☐ Universidad

☐ Diploma universitario ☐ Diploma de posgrado o superior

6. ¿Cuál es el nivel de educación superior que completó su pareja?

☐ Inferior al diploma de la escuela secundaria ☐ Diploma de la escuela secundaria ☐ Universidad

☐ Diploma universitario ☐ Diploma de posgrado o superior

7. ¿Cuál es su estado civil actual?

☐ Soltera ☐ Viuda ☐ En una relación estable

☐ Casada ☐ Divorciada ☐ Separada

8. ¿Cuántas veces estuvo embarazada en el pasado?

☐ 0 ☐ 1 ☐ 2 ☐ 3 o más

9. ¿Cuántos hijos tiene actualmente?

☐ 0 ☐ 1 ☐ 2 ☐ 3 o más

10. ¿Por qué motivo la visitó un asesor genético el día de hoy?

☐ Edad (mayor de 35) ☐ Resultado positivo del análisis de sangre/prueba de diagnóstico ☐ Otro: _____

11. ¿Decidió realizar una prueba genética después de la sesión de hoy?

☐ Sí ☐ No

11a. Si su respuesta a la pregunta anterior es **NO**, **PROCEDA** a la pregunta 12.

11b. Si su respuesta a la pregunta anterior es **SÍ**, ¿a qué prueba decidió someterse? (Marque todas las opciones que correspondan)

☐ Cariotipo en biopsia de vellosidades coriales (análisis de cromosomas)

☐ Amniocentesis para obtener el cariotipo (análisis de cromosomas)

☐ Biopsia de vellosidades coriales y estudio de microarray

☐ Amniocentesis y estudio de microarray

☐ Diagnóstico prenatal no invasivo

☐ Otro: _____

11c. Si la respuesta es **SÍ:** Para cada uno de los siguientes factores, encierre en un círculo un número entre el 1 y el 5 (1 = menos importante; 5 = más importante) para indicar cómo cada factor influenció su decisión con respecto a las pruebas. También, incluya los factores que cree que influenciaron más o menos a su pareja:

Encierre en un círculo un solo número, **tanto para usted como para su pareja,** para cada afirmación:

1 = menos importante **5 = más importante**

	Usted	**Su pareja**
Dolor de la prueba (biopsia de vellosidades coriales o amniocentesis):	1 2 3 4 5	1 2 3 4 5
Riesgo de la prueba (biopsia de vellosidades coriales o amniocentesis):	1 2 3 4 5	1 2 3 4 5
Preocupación por la salud del embarazo/bebé:	1 2 3 4 5	1 2 3 4 5
Antecedentes de embarazos previos:	1 2 3 4 5	1 2 3 4 5
Costo de la prueba o inquietudes con respecto al seguro:	1 2 3 4 5	1 2 3 4 5
Costos financieros de un niño con discapacidades:	1 2 3 4 5	1 2 3 4 5
Inquietud o demanda social de criar a un niño con discapacidades:	1 2 3 4 5	1 2 3 4 5
Limitaciones o incertidumbre con respecto a las pruebas genéticas:	1 2 3 4 5	1 2 3 4 5
Posibilidad de interrumpir el embarazo, tras un resultado anormal:	1 2 3 4 5	1 2 3 4 5

11d. ¿Qué nivel de influencia tuvieron los pensamientos, las inquietudes o creencias de su pareja, expresados en las afirmaciones anteriores, sobre su decisión de realizar las pruebas genéticas?

☐ No tuvieron influencia ☐ Influencia baja ☐ Influencia moderada ☐ Influencia alta

12. Si su respuesta a la pregunta 11 es **"NO"**. Para cada uno de los siguientes factores, encierre en un círculo un número entre el 1 y el 5 (1 = menos importante; 5 = más importante) para indicar cómo cada factor influenció su decisión de **NO** someterse a pruebas genéticas. También, incluya los factores que cree que influenciarían más o menos a su pareja:

	Usted	**Su pareja**
Encierre en un círculo un solo número, **tanto para usted como para su pareja,** para cada afirmación:	**1 = menos importante**	**5 = más importante**
Dolor de la prueba (biopsia de vellosidades coriales o amniocentesis):	1 2 3 4 5	1 2 3 4 5
Riesgo de la prueba (biopsia de vellosidades coriales o amniocentesis):	1 2 3 4 5	1 2 3 4 5
Preocupación por la salud del embarazo/bebé:	1 2 3 4 5	1 2 3 4 5
Antecedentes de embarazos previos:	1 2 3 4 5	1 2 3 4 5
Costo de la prueba o inquietudes con respecto al seguro:	1 2 3 4 5	1 2 3 4 5
Costos financieros de un niño con discapacidades:	1 2 3 4 5	1 2 3 4 5
Inquietud o demanda social de criar a un niño con discapacidades:	1 2 3 4 5	1 2 3 4 5
Limitaciones o incertidumbre con respecto a las pruebas genéticas:	1 2 3 4 5	1 2 3 4 5
Posibilidad de interrumpir el embarazo, tras un resultado anormal:	1 2 3 4 5	1 2 3 4 5

12a. Si su respuesta a la pregunta 11 es **"NO"**. ¿Qué nivel de influencia tuvieron los pensamientos, las inquietudes o creencias de su pareja, expresados en las afirmaciones anteriores, sobre su decisión de no realizar las pruebas genéticas?

☐ No tuvieron influencia ☐ Influencia baja ☐ Influencia moderada ☐ Influencia alta

12b. ¿Hubiese considerado la opción de someterse a pruebas genéticas, si su pareja **la hubiese acompañado** en la sesión de asesoría genética? Si la respuesta es "sí", **seleccione todas** las pruebas con cuya realización se hubiese sentido cómoda si su pareja hubiese estado presente:

☐ Cariotipo en biopsia de vellosidades coriales (análisis de cromosomas)

☐ Amniocentesis para obtener el cariotipo (análisis de cromosomas)

☐ Biopsia de vellosidades coriales y estudio de microarray

☐ Amniocentesis y estudio de microarray

☐ Diagnóstico prenatal no invasivo

☐ Otro: _____

☐ Ninguna prueba

13. ¿Por qué su pareja no estuvo presente en la sesión?

☐ Por trabajo ☐ Por una enfermedad o lesión

☐ Estamos separados/no participará en la vida del bebé ☐ Otro: _____

14. ¿Cree que hubiese sido útil que su pareja esté en la sesión?

☐ No ☐ Muy poco útil ☐ Algo útil ☐ Muy útil

15. ¿Usted o su pareja, por lo general, toman las decisiones durante el embarazo?

☐ Mi pareja ☐ Yo ☐ Compartimos las decisiones en igual medida

16. ¿Su pareja participó de forma significativa en las decisiones con respecto a embarazos anteriores? Por ejemplo, dónde recibir atención durante el embarazo, tipo de parto deseado, medicamentos, o decisiones ante complicaciones, etc.

☐ Para nada ☐ Casi no participó ☐ Participó un poco ☐ Participó mucho ☐ No corresponde

17. ¿Quién tiende a tomar la mayoría de las decisiones con respecto a dónde comer, dónde vivir, cómo pasar el tiempo libre juntos, etc.?

☐ Siempre yo ☐ Por lo general yo ☐ Los dos por igual ☐ Por lo general mi pareja ☐ Siempre mi pareja

18. ¿A quién considera parte de su red de apoyo a la hora de tomar decisiones difíciles?
(seleccione todas las opciones que correspondan)

☐ Pareja ☐ Familia

☐ Amigos ☐ Proveedor de atención

☐ Iglesia/fe ☐ Mascotas

☐ Medios de comunicación social/ televisión/internet ☐ Otro: _____

APPENDIX N

Questionnaire for Mother with Partner Present

Purpose: In this study, we hope to find out more about how you feel about genetic testing. This study will explore how people make choices about genetic testing during pregnancy. Participating in this study is voluntary and requires you to answer all of the questions below. Please answer all questions individually without your partner.

You may ask the researchers any questions that come up while taking the survey. If any questions should arise, feel free to contact the researcher using the contact information below:

Dillon van den Berg, Lead Researcher
University of California, Irvine
(714) 456-5837 or dvandenb@uci.edu

Confidentiality: Any information provided on this questionnaire will remain confidential. Only the researchers will have access to the questionnaires and the information on it. No identifying information will be included on the survey and any personal information will not be revealed. The results of this research may be published in a scientific journal and/or presented at a professional meeting.

Please mark each question with a check in the appropriate box or fill in the blank if prompted.

1. Your Age Group:

☐ Under 25 ☐ 25-35 ☐ 36-45

☐ 46-55 ☐ 55+

2. Which ethnic group describes you the best? (Please check only one)

☐ African American/Black ☐ White/Caucasian

☐ American Indian/Alaskan Native ☐ Hispanic/Latino

☐ Asian/Pacific Islander ☐ Other: (please specify) _____

3. What is the highest level of education that you have completed?

☐ Less than a High School degree ☐ High school degree ☐ Some college

☐ College degree ☐ Postgraduate degree or higher

4. What is your current relationship/marital status?

☐ Single ☐ Widowed ☐ Committed relationship

☐ Married ☐ Divorced ☐ Separated

5. How many times have you been pregnant previously?

☐ 0 ☐ 1 ☐ 2 ☐ 3 or more

6. How many children do you currently have?

☐ 0 ☐ 1 ☐ 2 ☐ 3 or more

7. For what reason were you and your partner seen by a Genetic Counselor today?

☐ Age (35+) ☐ Positive blood test ☐ Other: _____

8. Did you decide to have genetic testing after today's session?

☐ Yes ☐ No

8a. If **NO** to the above question, **SKIP** to Question #9.

8b. If **YES** to above, which testing did you decide to have (check all that apply)?

☐ CVS with Karyotype (chromosome analysis)
☐ Amniocentesis with Karyotype (chromosome analysis)
☐ CVS with Microarray
☐ Amniocentesis with Microarray
☐ Noninvasive prenatal screening (NIPT)
☐ Other: _____

8c. If **you decide to have the test:** For each of the following factors, circle a number between 1 and 5 (1=least important & 5=very important) for how important each factor was in your testing decision. Also circle a number for how important you believe each factor was for your partner:

Circle only one number for both you and your partner for each statement:	**You** 1=least important	**Your Partner** 5=most important
Pain of the test (CVS or Amniocentesis):	1 2 3 4 5	1 2 3 4 5
Risk of the test (CVS or Amniocentesis):	1 2 3 4 5	1 2 3 4 5
Concern about health of pregnancy/baby:	1 2 3 4 5	1 2 3 4 5
Past pregnancy history:	1 2 3 4 5	1 2 3 4 5
Cost of testing or insurance worries:	1 2 3 4 5	1 2 3 4 5
Financial burden of a child with disabilities:	1 2 3 4 5	1 2 3 4 5
Social issue/demand of raising a child with disabilities:	1 2 3 4 5	1 2 3 4 5
Limitations or uncertainty about genetic testing:	1 2 3 4 5	1 2 3 4 5
Option to stop the pregnancy if the result is abnormal:	1 2 3 4 5	1 2 3 4 5

8d. How much of an influence did your partner's thoughts, concerns or beliefs from the above statements have on your decision to pursue testing?

☐ No influence ☐ Low influence ☐ Moderate Influence ☐ High influence

8e. Would you have still considered genetic testing, if your partner was **not** present at the genetic counseling session with you? If yes, select all tests that you would have felt comfortable making without your partner present:

☐ CVS with Karyotype (chromosome analysis)
☐ Amniocentesis with Karyotype (chromosome analysis)
☐ CVS with Microarray
☐ Amniocentesis with Microarray

☐ Noninvasive prenatal screening (NIPT) ☐ Other: _____

☐ No Testing

9. If you decided NOT to have a test and answered **"NO"** to question #8: For each of the following factors, circle a number between 1 and 5 (1=least important & 5=most important) for how important each factor was in your decision **NOT** to have genetic testing. Also circle a number for how important you believe each factor was for your partner:

	You	**Your Partner**
Circle only one number for both you and your partner for each statement:	**1=least important**	**5=most important**
Pain of the test (CVS or Amniocentesis):	1 2 3 4 5	1 2 3 4 5
Risk of the test (CVS or Amniocentesis):	1 2 3 4 5	1 2 3 4 5
Concern about health of pregnancy/baby:	1 2 3 4 5	1 2 3 4 5
Past pregnancy history:	1 2 3 4 5	1 2 3 4 5
Cost of testing or insurance worries:	1 2 3 4 5	1 2 3 4 5
Financial burden of a child with disabilities:	1 2 3 4 5	1 2 3 4 5
Social issue/demand of raising a child with disabilities:	1 2 3 4 5	1 2 3 4 5
Limitations or uncertainty about genetic testing:	1 2 3 4 5	1 2 3 4 5
Option to stop the pregnancy if the result is abnormal:	1 2 3 4 5	1 2 3 4 5

9a. If you answered **"NO"** to question #8: How much of an influence did your partner's thoughts, concerns or beliefs from the above statements have on your decision to **NOT** pursue testing?

☐ No influence ☐ Low influence ☐ Moderate Influence ☐ High influence

10. Does either you or your partner usually take the lead in decisions during this pregnancy?

☐ My partner does ☐ I take the lead ☐ We share decisions equally

11. Was it helpful to have your partner present in the session?

☐ Not helpful ☐ A little helpful ☐ Moderately Helpful ☐ Very helpful

12. Was your partner heavily involved in decisions with past pregnancies, such as: where you went for pregnancy care, type of delivery desired, medication taken, or decisions made when complications have arisen, etc.?

☐ Not involved ☐ A little involved ☐ Moderately involved ☐ Very involved ☐ N/A

13. Who in the relationship tends to be the primary decision maker regarding: where to eat, where to live, how to spend free time together, etc.?

☐ Always Me ☐ Usually Me ☐ Equal ☐ Usually my partner ☐ Always my partner

13. Whom do you consider to be part of your support network in making difficult decisions? (select all that apply)

- ☐ Partner
- ☐ Family
- ☐ Friends
- ☐ Doctor, nurse or other care provider
- ☐ Church/Faith
- ☐ Pets
- ☐ Social Media/TV/Internet
- ☐ Other: _____

APPENDIX O

Cuestionario para la Madre Con la Pareja Presente

Objetivo: En este estudio, deseamos saber más acerca de cómo se siente con respecto a las pruebas genéticas. Exploraremos la forma en que las personas toman decisiones con respecto a las pruebas genéticas durante el embarazo. La participación en este estudio es voluntaria y requiere que responda todas las preguntas que se encuentran a continuación. Responda todas las preguntas de forma individual, sin su pareja.

Puede hacerles todas las preguntas que surjan durante la encuesta a los investigadores. Si surgen preguntas, no dude en comunicarse con el investigador, utilizando la siguiente información de contacto:

Dillon van den Berg, investigador principal
University of California, Irvine
(714) 456-5837 o dvandenb@uci.edu

Confidencialidad: Toda la información proporcionada en este cuestionario será confidencial. Solo los investigadores tendrán acceso a los cuestionarios y a la información contenida en ellos. No se incluirá información de identificación en la encuesta, ni se revelará información personal. Es posible que los resultados de esta investigación se publiquen en una revista científica o se presenten en una reunión profesional.

Marque la casilla correspondiente a cada pregunta o complete el espacio cuando se solicita.

1. Su grupo etario:

☐ Menor de 25 ☐ 25-35 ☐ 36-45

☐ 46-55 ☐ Mayor de 55

2. ¿Con qué grupo étnico se identifica más? (Marque solo una opción)

☐ Afroamericano/negro ☐ Blanco/caucásico ☐ Amerindio/nativo de Alaska

☐ Hispano/latino ☐ Asiático/isleño del Pacífico ☐ Otro (especifique): _____

3. ¿Cuál es el nivel de educación superior que completó?

☐ Inferior al diploma de la escuela secundaria ☐ Diploma de la escuela secundaria ☐ Universidad

☐ Diploma universitario ☐ Diploma de posgrado o superior

4. ¿Cuál es su estado civil actual?

☐ Soltera ☐ Viuda ☐ En una relación estable

☐ Casada ☐ Divorciada ☐ Separada

5. ¿Cuántas veces estuvo embarazada en el pasado?

☐ 0 ☐ 1 ☐ 2 ☐ 3 o más

6. ¿Cuántos hijos tiene actualmente?

☐ 0 ☐ 1 ☐ 2 ☐ 3 o más

7. ¿Por qué motivo los visitó un asesor genético a usted y a su pareja el día de hoy?

☐ Edad (mayor de 35) ☐ Resultado positivo del análisis de sangre ☐ Otro: _____

8. ¿Decidió realizar una prueba genética después de la sesión de hoy?

☐ Sí ☐ No

8a. Si su respuesta a la pregunta anterior es **NO**, **PROCEDA** a la pregunta 9.

8b. Si su respuesta a la pregunta anterior es **SÍ**, ¿a qué prueba decidió someterse? (Marque todas las opciones que correspondan)

☐ Cariotipo en biopsia de vellosidades coriales (análisis de cromosomas)

☐ Amniocentesis para obtener el cariotipo (análisis de cromosomas)

☐ Biopsia de vellosidades coriales y estudio de microarray

☐ Amniocentesis y estudio de microarray

☐ Diagnóstico prenatal no invasivo

☐ Otro: _____

8c. Si la respuesta es **SÍ:** Para cada uno de los siguientes factores, encierre en un círculo un número entre el 1 y el 5 (1 = menos importante; 5 = más importante) para indicar cómo cada factor influenció su decisión con respecto a las pruebas. También, incluya los factores que cree que influenciarían más o menos a su pareja a la hora de elegir la prueba, **sin preguntarle a su pareja**:

Encierre en un círculo un solo número, **tanto para usted como para su pareja,** para cada afirmación:

1 = menos importante **5 = más importante**

	Usted	**Su pareja**
Dolor de la prueba (biopsia de vellosidades coriales o amniocentesis):	1 2 3 4 5	1 2 3 4 5
Riesgo de la prueba (biopsia de vellosidades coriales o amniocentesis):	1 2 3 4 5	1 2 3 4 5
Preocupación por la salud del embarazo/bebé:	1 2 3 4 5	1 2 3 4 5
Antecedentes de embarazos previos:	1 2 3 4 5	1 2 3 4 5
Costo de la prueba o inquietudes con respecto al seguro:	1 2 3 4 5	1 2 3 4 5
Costos financieros de un niño con discapacidades:	1 2 3 4 5	1 2 3 4 5
Inquietud o demanda social de criar a un niño con discapacidades:	1 2 3 4 5	1 2 3 4 5
Limitaciones o incertidumbre con respecto a las pruebas genéticas:	1 2 3 4 5	1 2 3 4 5
Posibilidad de interrumpir el embarazo, tras un resultado anormal:	1 2 3 4 5	1 2 3 4 5

8d. ¿Qué nivel de influencia tuvieron los pensamientos, las inquietudes o creencias de su pareja, expresados en las afirmaciones anteriores, sobre su decisión de realizar las pruebas genéticas?

☐ No tuvieron influencia ☐ Influencia baja ☐ Influencia moderada ☐ Influencia alta

8e. ¿Hubiese considerado la opción de someterse a pruebas genéticas, si su pareja **no** la hubiese acompañado en la sesión de asesoría genética? Si la respuesta es "sí", seleccione todas las pruebas con cuya realización se hubiese sentido cómoda si su pareja no hubiese estado presente:

☐ Cariotipo en biopsia de vellosidades coriales (análisis de cromosomas)

☐ Amniocentesis para obtener el cariotipo (análisis de cromosomas)

☐ Biopsia de vellosidades coriales y estudio de microarray

☐ Amniocentesis y estudio de microarray

☐ Diagnóstico prenatal no invasivo ☐ Otro: _____

☐ Ninguna prueba

9. Si su respuesta a la pregunta 8 es **"NO"**. Para cada uno de los siguientes factores, encierre en un círculo un número entre el 1 y el 5 (1 = menos importante; 5 = más importante) para indicar cómo cada factor influenció su decisión de **NO** someterse a pruebas genéticas. También, incluya los factores que cree que influenciaron más o menos a su pareja a la hora de elegir NO realizar la prueba, **sin preguntarle a su pareja**:

	Usted	**Su pareja**
Encierre en un círculo un solo número, **tanto para usted como para su pareja,** para cada afirmación:	**1 = menos importante** **5 = más importante**	
Dolor de la prueba (biopsia de vellosidades coriales o amniocentesis):	1 2 3 4 5	1 2 3 4 5
Riesgo de la prueba (biopsia de vellosidades coriales o amniocentesis):	1 2 3 4 5	1 2 3 4 5
Preocupación por la salud del embarazo/bebé:	1 2 3 4 5	1 2 3 4 5
Antecedentes de embarazos previos:	1 2 3 4 5	1 2 3 4 5
Costo de la prueba o inquietudes con respecto al seguro:	1 2 3 4 5	1 2 3 4 5
Costos financieros de un niño con discapacidades:	1 2 3 4 5	1 2 3 4 5
Inquietud o demanda social de criar a un niño con discapacidades:	1 2 3 4 5	1 2 3 4 5
Limitaciones o incertidumbre con respecto a las pruebas genéticas:	1 2 3 4 5	1 2 3 4 5
Posibilidad de interrumpir el embarazo, tras un resultado anormal:	1 2 3 4 5	1 2 3 4 5

9a. ¿Qué nivel de influencia tuvieron los pensamientos, las inquietudes o creencias de su pareja, expresados en las afirmaciones anteriores, sobre su decisión de **NO** realizar las pruebas genéticas

☐ No tuvieron influencia ☐ Influencia baja ☐ Influencia moderada ☐ Influencia alta

10. ¿Usted o su pareja, por lo general, toman las decisiones durante el embarazo?

☐ Mi pareja ☐ Yo ☐ Compartimos las decisiones en igual medida

11. ¿Cree que fue útil que su pareja esté presente en la sesión?

☐ No ☐ Muy poco útil ☐ Algo útil ☐ Muy útil

12. ¿Su pareja participó de forma significativa en las decisiones con respecto a embarazos anteriores? Por ejemplo, dónde recibir atención durante el embarazo, tipo de parto deseado, medicamentos, o decisiones ante complicaciones, etc.

☐ Para nada ☐ Casi no participó ☐ Participó un poco ☐ Participó mucho ☐ No corresponde

13. ¿Quién tiende a tomar la mayoría de las decisiones con respecto a dónde comer, dónde vivir, cómo pasar el tiempo libre juntos, etc.?

☐ Siempre yo ☐ Por lo general yo ☐ Los dos por igual ☐ Por lo general mi pareja ☐ Siempre mi pareja

14. ¿A quién considera parte de su red de apoyo a la hora de tomar decisiones difíciles? (seleccione todas las opciones que correspondan)

- ☐ Pareja
- ☐ Familia
- ☐ Amigos
- ☐ Proveedor de atención
- ☐ Iglesia/fe
- ☐ Mascotas
- ☐ Medios de comunicación social/ televisión/internet
- ☐ Otro: _____

APPENDIX P

Questionnaire for Partner

Purpose: In this study, we hope to find out more about how you feel about genetic testing. This study will explore how people make choices about genetic testing during pregnancy. Participating in this study is voluntary and requires you to answer all of the questions below. Please answer all questions on your own without your partner.

You may ask the researchers any questions that come up while taking the survey. If you have any questions, feel free to contact the researcher using the contact information below:

Dillon van den Berg, Lead Researcher
University of California, Irvine
(714) 456-5837 or dvandenb@uci.edu

Confidentiality: Any information provided on this questionnaire will remain confidential. Only the researchers will have access to the questionnaires and the information on it. No identifying information will be included on the survey and any personal information will not be revealed. The results of this research may be published in a scientific journal and/or presented at a professional meeting.

Please mark each question with a check in the appropriate box or fill in the blank if asked to.

1. Your Age Group:

☐ Under 25 ☐ 25-35 ☐ 36-45

☐ 46-55 ☐ 55+

2. Which ethnic group describes you the best? (Please check only one)

☐ African American/Black ☐ White/Caucasian

☐ American Indian/Alaskan Native ☐ Hispanic/Latino

☐ Asian/Pacific Islander ☐ Other: (please specify) _____

3. What is the highest level of education that you have completed?

☐ Less than a High School degree ☐ High school degree ☐ Some college

☐ College degree ☐ Postgraduate degree or higher

4. What is your current relationship/marital status?

☐ Single ☐ Widowed ☐ Committed relationship

☐ Married ☐ Divorced ☐ Separated

5. How many children do you currently have? (including past relationships)

☐ 0 ☐ 1

☐ 2 ☐ 3 or more

6. For what reason were you and your partner seen by a Genetic Counselor today?

☐ Age (35+) ☐ Positive blood test ☐ Other: _____

7. Did you and your partner decide to have genetic testing after today's session?

☐ Yes ☐ No

7a. If **NO** to the above question, **SKIP** to Question #8.

7b. If "**YES**" to above, which testing did you and your partner elect to have (check all that apply)?

☐ CVS with Karyotype (chromosome analysis)

☐ Amniocentesis with Karyotype (chromosome analysis)

☐ CVS with Microarray

☐ Amniocentesis with Microarray

☐ Noninvasive prenatal screening (NIPT)

☐ Other: _____

7c. If **you decide to have the test:** For each of the following factors, circle a number between 1 and 5 (1=least important & 5=very important) for how important each factor was in your testing decision. Also circle a number for how important you believe each factor was for your partner.

Circle only one number for both you and your partner for each statement:	**You** **1=least important**	**Your Partner** **5=most important**
Pain from the test (CVS or Amniocentesis):	1 2 3 4 5	1 2 3 4 5
Risk from the test (CVS or Amniocentesis):	1 2 3 4 5	1 2 3 4 5
Concern about health of pregnancy/baby:	1 2 3 4 5	1 2 3 4 5
Past pregnancy history:	1 2 3 4 5	1 2 3 4 5
Cost of testing or insurance worries:	1 2 3 4 5	1 2 3 4 5
Financial burden of a child with disabilities:	1 2 3 4 5	1 2 3 4 5
Social issue/difficulty of raising a child with disabilities:	1 2 3 4 5	1 2 3 4 5
Limitations or uncertainty about genetic testing:	1 2 3 4 5	1 2 3 4 5
Option of stopping pregnancy if result is abnormal:	1 2 3 4 5	1 2 3 4 5

7d. How much of an influence did your thoughts, concerns or beliefs from the above statements have on your decision to pursue testing?

☐ No influence ☐ Low influence ☐ Moderate Influence ☐ High influence

8. If you decided NOT to have a test and answered **"NO"** to question #7: For each of the following factors, circle a number between 1 and 5 (1=least important & 5=most important) for how important each factor was in your decision **NOT** to have genetic testing. Also circle a number for how important you believe each factor was for your partner.

	You	**Your Partner**
Circle only one number for both you and your partner for each statement:	**1=least important**	**5=most important**
Pain from the test (CVS or Amniocentesis):	1 2 3 4 5	1 2 3 4 5
Risk from the test (CVS or Amniocentesis):	1 2 3 4 5	1 2 3 4 5
Concern about health of pregnancy/baby:	1 2 3 4 5	1 2 3 4 5
Past pregnancy history:	1 2 3 4 5	1 2 3 4 5
Cost of testing or insurance worries:	1 2 3 4 5	1 2 3 4 5
Financial burden of a child with disabilities:	1 2 3 4 5	1 2 3 4 5
Social issue/difficulty of raising a child with disabilities:	1 2 3 4 5	1 2 3 4 5
Limitations or uncertainty about genetic testing:	1 2 3 4 5	1 2 3 4 5
Option of stopping pregnancy if result is abnormal:	1 2 3 4 5	1 2 3 4 5

8a. If you answered **"NO"** to question #7: How much of an influence did your thoughts, concerns or beliefs from the above statements have on your decision to **NOT** pursue testing?

☐ No influence ☐ Low influence ☐ Moderate Influence ☐ High influence

9. Does either you or your partner usually take the lead in decisions during this pregnancy?

☐ My partner does ☐ I take the lead ☐ We share decisions equally

10. Do you think it was helpful to your partner for you to be present at the session?

☐ Not helpful ☐ A little helpful ☐ Moderately helpful ☐ Very helpful

11. How involved have you been in decisions with past pregnancies, such as: where you went for pregnancy care, type of delivery desired, medication taken, or decisions made when complications have arisen, etc?

☐ Not involved ☐ A little involved ☐ Moderately involved ☐ Very involved ☐ N/A

12. Who in the relationship tends to be the primary decision maker regarding: where to eat, where to live, how to spend free time together, etc.?

☐ Always Me ☐ Usually Me ☐ Equal ☐ Usually my partner ☐ Always my partner

13. Whom do you consider to be part of your support network in making difficult decisions? (select all that apply)

- ☐ Partner
- ☐ Family
- ☐ Friends
- ☐ Doctor, nurse or other care provider
- ☐ Church/Faith
- ☐ Pets
- ☐ Social Media/TV/Internet
- ☐ Other: _____

APPENDIX Q

Cuestionario para la Pareja

Objetivo: En este estudio, deseamos saber más acerca de cómo se siente con respecto a las pruebas genéticas. Exploraremos la forma en que las personas toman decisiones con respecto a las pruebas genéticas durante el embarazo. La participación en este estudio es voluntaria y requiere que responda todas las preguntas que se encuentran a continuación. Responda todas las preguntas de forma individual, sin su pareja.

Puede hacerles todas las preguntas que surjan durante la encuesta a los investigadores. Si surgen preguntas, no dude en comunicarse con el investigador, utilizando la siguiente información de contacto:

Dillon van den Berg, investigador principal
University of California, Irvine
(714) 456-5837 o dvandenb@uci.edu

Confidencialidad: Toda la información proporcionada en este cuestionario será confidencial. Solo los investigadores tendrán acceso a los cuestionarios y a la información contenida en ellos. No se incluirá información de identificación en la encuesta, ni se revelará información personal. Es posible que los resultados de esta investigación se publiquen en una revista científica o se presenten en una reunión profesional.

Marque la casilla correspondiente a cada pregunta o complete el espacio cuando se solicita.

1. Su grupo etario:

☐ Menor de 25 ☐ 25-35 ☐ 36-45

☐ 46-55 ☐ Mayor de 55

2. ¿Con qué grupo étnico se identifica más? (Marque solo una opción)

☐ Afroamericano/negro ☐ Blanco/caucásico

☐ Amerindio/nativo de Alaska ☐ Hispano/latino

☐ Asiático/isleño del Pacífico ☐ Otro (especifique): _____

3. ¿Cuál es el nivel de educación superior que completó?

☐ Inferior al diploma de la escuela secundaria ☐ Diploma de la escuela secundaria ☐ Universidad

☐ Diploma universitario ☐ Diploma de posgrado o superior

4. ¿Cuál es su estado civil actual?

☐ Soltero/a ☐ Viudo/a ☐ En una relación estable

☐ Casada ☐ Divorciado/a ☐ Separado/a

5. ¿Cuántos hijos tiene actualmente? (incluidos los hijos de relaciones anteriores)

☐ 0 ☐ 1 ☐ 2 ☐ 3 o más

6. ¿Por qué motivo los visitó un asesor genético a usted y a su pareja el día de hoy?

☐ Edad (mayor de 35) ☐ Resultado positivo del análisis de sangre ☐ Otro: _____

7. ¿Usted y su pareja decidieron realizar una prueba genética después de la sesión de hoy?

☐ Sí ☐ No

7a. Si su respuesta a la pregunta anterior es **NO**, **PROCEDA** a la pregunta 8.

7b. Si su respuesta a la pregunta anterior es **SÍ**, ¿a qué prueba decidió someterse? (Marque todas las opciones que correspondan)

☐ Cariotipo en biopsia de vellosidades coriales (análisis de cromosomas)

☐ Amniocentesis para obtener el cariotipo (análisis de cromosomas)

☐ Biopsia de vellosidades coriales y estudio de microarray

☐ Amniocentesis y estudio de microarray

☐ Diagnóstico prenatal no invasivo

☐ Otro: _____

7c. Si la respuesta es **SÍ:** Para cada uno de los siguientes factores, encierre en un círculo un número entre el 1 y el 5 (1 = menos importante; 5 = más importante) para indicar cómo cada factor influenció su decisión con respecto a las pruebas. También, incluya los factores que cree que influenciarían más o menos a su pareja a la hora de elegir la prueba, **sin preguntarle a su pareja**:

Encierre en un círculo un solo número, **tanto para usted como para su pareja,** para cada afirmación:

	Usted (**1 = menos importante** … **5 = más importante**)	**Su pareja** (**1 = menos importante** … **5 = más importante**)
Dolor la prueba (biopsia de vellosidades coriales o amniocentesis):	1 2 3 4 5	1 2 3 4 5
Riesgo la prueba (biopsia de vellosidades coriales o amniocentesis):	1 2 3 4 5	1 2 3 4 5
Preocupación por la salud del embarazo/bebé:	1 2 3 4 5	1 2 3 4 5
Antecedentes de embarazos previos:	1 2 3 4 5	1 2 3 4 5
Costo de la prueba o inquietudes con respecto al seguro:	1 2 3 4 5	1 2 3 4 5
Costos financieros de un niño con discapacidades:	1 2 3 4 5	1 2 3 4 5
Inquietud o demanda social de criar a un niño con discapacidades:	1 2 3 4 5	1 2 3 4 5
Limitaciones o incertidumbre con respecto a las pruebas genéticas:	1 2 3 4 5	1 2 3 4 5
Posibilidad de interrumpir el embarazo, tras un resultado anormal:	1 2 3 4 5	1 2 3 4 5

7d. ¿Qué nivel de influencia tuvieron sus pensamientos, inquietudes o creencias, expresados en las afirmaciones anteriores, sobre su decisión de realizar las pruebas genéticas, después de la sesión de hoy?

☐ No tuvieron influencia ☐ Influencia baja ☐ Influencia moderada ☐ Influencia alta

8. Si su respuesta a la pregunta 8 es **"NO"**. Para cada uno de los siguientes factores, encierre en un círculo un número entre el 1 y el 5 (1 = menos importante; 5 = más importante) para indicar cómo cada factor influenció su decisión de **NO** someterse a pruebas genéticas. También, incluya los factores que cree que influenciaron más o menos a su pareja a la hora de elegir NO realizar la prueba, **sin preguntarle a su pareja**:

Encierre en un círculo un solo número, **tanto para usted como para su pareja,** para cada afirmación:

	Usted (**1 = menos importante**)	**Su pareja** (**5 = más importante**)
Dolor de la prueba (biopsia de vellosidades coriales o amniocentesis):	1 2 3 4 5	1 2 3 4 5
Riesgo de la prueba (biopsia de vellosidades coriales o amniocentesis):	1 2 3 4 5	1 2 3 4 5
Preocupación por la salud del embarazo/bebé:	1 2 3 4 5	1 2 3 4 5
Antecedentes de embarazos previos:	1 2 3 4 5	1 2 3 4 5
Costo de la prueba o inquietudes con respecto al seguro:	1 2 3 4 5	1 2 3 4 5
Costos financieros de un niño con discapacidades:	1 2 3 4 5	1 2 3 4 5
Inquietud o demanda social de criar a un niño con discapacidades:	1 2 3 4 5	1 2 3 4 5
Limitaciones o incertidumbre con respecto a las pruebas genéticas:	1 2 3 4 5	1 2 3 4 5
Posibilidad de interrumpir el embarazo, tras un resultado anormal:	1 2 3 4 5	1 2 3 4 5

8a. ¿Qué nivel de influencia tuvieron sus pensamientos, inquietudes o creencias, expresados en las afirmaciones anteriores, sobre su decisión de **NO** realizar las pruebas genéticas?

☐ No tuvieron influencia ☐ Influencia baja ☐ Influencia moderada ☐ Influencia alta

9. ¿Usted o su pareja, por lo general, toman las decisiones durante el embarazo?

☐ Mi pareja ☐ Yo ☐ Compartimos las decisiones en igual medida

10. ¿Cree que fue útil para su pareja que usted esté presente en la sesión?

☐ Para nada útil ☐ Muy poco útil ☐ Algo útil ☐ Muy útil

11. ¿Siente que participó de forma significativa en las decisiones con respecto a embarazos anteriores? Por ejemplo, dónde recibir atención durante el embarazo, tipo de parto deseado, medicamentos, o decisiones ante complicaciones, etc.

☐ Para nada ☐ Casi no participó ☐ Participó un poco ☐ Participó mucho ☐ No corresponde

12. ¿Quién tiende a tomar la mayoría de las decisiones con respecto a dónde comer, dónde vivir, cómo pasar el tiempo libre juntos, etc.?

☐ Siempre yo ☐ Por lo general yo ☐ Los dos por igual ☐ Por lo general mi pareja ☐ Siempre mi pareja

13. ¿A quién considera parte de su red de apoyo a la hora de tomar decisiones difíciles? (seleccione todas las opciones que correspondan)

- ☐ Pareja
- ☐ Familia
- ☐ Amigos
- ☐ Proveedor de atención
- ☐ Iglesia/fe
- ☐ Mascotas
- ☐ Medios de comunicación social/ televisión/internet
- ☐ Otro: _____