Title

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"It Became Easier Once I Knew": Stakeholder Perspectives for Educating Children and Teenagers about Their Difference of Sex Development

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We confirm all patient/personal identifiers have been removed or disguised so the patient/person(s) described are not identifiable and cannot be identified through the details of the manuscript.

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Abstract

Objective: Secrecy about a child's difference of sex development (DSD) can lead to internalized shame and stigma. We explored how teenagers and adults with DSD, parents, healthcare providers, and allied professionals value and perceive patient education.

Methods: Stakeholders (n = 110) completed qualitative semi-structured interviews. Relevant themes for educational content were queried and organized.

Results: Education was consistently identified as essential to successful outcomes. There was less consistency in *how* to educate patients. Disagreement existed regarding *who* should champion the education process. Participants believed medically relevant information should be shared gradually with attention to developmental capacity. Details were lacking regarding *how* much or *what* information to share. Participants noted that vetted resources were helpful. Benefits of sharing condition-specific information with patients included supporting their psychosocial development. Barriers included parental resistance to sharing information due to shame/stigma, and cultural and/or family dynamics.

Conclusions: Stakeholders' different perspectives regarding patient DSD education warrant future research to focus on the design, evaluation, and implementation of education-focused interventions.

Keywords

differences of sex development; disorders of sex development; intersex; education; disclosure

1. Introduction

Fully informing children of their diagnoses was not always standard practice in medicine. Before the 1960's, healthcare providers would often shield children from knowing about or fully understanding chronic or disabling medical conditions. It was believed that children could not comprehend the complexities and implications of a diagnosis; therefore, it was best to protect them by withholding information.[1–3] In response to calls for greater patient autonomy and promotion of shared decision-making between providers and patients, the American Medical Association adopted a stance for full disclosure[1] and the American Academy of Pediatrics advocated for providing patients with information and education about their diagnoses.[4–6] The Institute of Medicine, discussed the continued need for patient-centeredness, advocating for shared decision making.[7] Ensuring children know their correct diagnosis and have accurate knowledge of their condition as they mature can promote adherence to medical treatments, assist in transition to adult care facilities, and

[1]Throughout this article, we opt to utilize the word "education" rather than "disclosure" to model for readers that education may be a preferred alternative due to the negative connotation that can be associated with disclosure (i.e., making known something that was previously secret or private).

enhance long-term psychosocial outcomes.[8–10] The withholding of information about a diagnosis leads to patients experiencing internalized shame and stigma, as well as mistrust of the medical community and their parents.[11–13]

Differences of sex development (DSD)[2] are congenital conditions where anatomic, genetic, or chromosomal sex development is atypical.[14] The range of DSD conditions are categorized as sex chromosome DSDs (e.g., Turner or Klinefelter syndrome), 46,XY DSDs (e.g., 5-alpha reductase deficiency or complete androgen insensitivity syndrome), and 46,XX DSDs (e.g., congenital adrenal hyperplasia). More common phenotypes include infertility, genital ambiguity, undescended testicles, gynecomastia in males, and virilization during puberty in females.[14] Despite the range of conditions and phenotypes, the parallels across DSD warrant similar considerations related to patient education. Within DSD healthcare, the preference for secrecy and withholding medical information from younger patients was common practice in the past.[15–17] Though not an official policy or directive, older literature held positions that educating patients about their diagnosis was discouraged.[18, 19] Personal accounts and other sources state that patients did not know about their diagnosis until they were much older, often not until adulthood. Providers would make patient care decisions to “fix” the atypical anatomy and parents were advised to withhold information about the DSD, surgical history, or treatment from their child for fear of shame or stigma.[2] The 2006 Consensus Statement and 2016 Update also encouraged providers to openly engage parents and patients in discussions, particularly surrounding the diagnosis and associated care.[1, 14] Further, there was a push for studies to evaluate timing and content of information provided to patients.[14] The 2016 Consensus Statement Update reiterated the importance of education from providers to patients through shared decision making and opening lines of communication regarding diagnosis, and recommended e-learning and scripts to assist the providers in sharing information with patients and families.[1] Integrating adult and patient advocates in DSD clinics may also support ongoing education of patients and families.

Even with multiple agencies and practice guidelines advocating for open communication and patient education within pediatric DSD care, studies on implementation and outcomes are lacking. Two papers that evaluated the education process in girls with androgen insensitivity syndrome and Turner syndrome recommended education of patients in an age-appropriate way.[20, 21] Further, the authors found some individuals experienced negative outcomes when parents were secretive about their condition. Older patients often vocalized having felt in the dark with regards to their diagnosis and past treatments. This secrecy leads to poor understanding of their condition, which could have implications for future care, and may result in internalized stigma and shame.[16, 22]

Within the broader pediatric literature, although there is support from the medical and psychological community to provide open education, some parents continue to attempt to protect their child from perceived difficult discussions or potential stigma by withholding

[2]The term “disorders of sex development” was proposed during the 2006 Consensus Statement on Intersex Conditions (Lee et al., 2006). However, the word “disorder” is considered by some to be stigmatizing. As such, we use the term “differences of sex development” to recognize the controversy over labels given to these conditions. Although “Intersex” is another term preferred by some, we opted to use other terms to promote person-first language.

medical information.[5, 6] In pediatric cancer, 30% of parents rated the presence of their child during a discussion regarding treatment and prognosis for a leukemia diagnosis as unfavorable and thought this negatively impacted conversations with providers; only 10% thought that having the child in the room was desirable.[23] However, research in pediatric cancer, pediatric HIV, and children born via egg donation has shown that educating children in a developmentally appropriate way can be beneficial.[24–26] When educated about their condition, pediatric patients with HIV are more likely to understand the full impact of their diagnosis (e.g., the potentially serious or even fatal implications of not taking medication). They also develop skills to ask appropriate questions regarding their illness and long-term care. Moreover, children and adolescents with cancer prefer to participate in decision-making and most (75%) believe it is appropriate to include them in end-of-life decisions.[26, 27] Some suggest that the child should guide the conversation on what information to provide and when.[28] The American Academy of Pediatrics recommends patient education to promote patient autonomy and child assent for medical decisions.[29]

Due to the limited findings within DSD, and potential implications in the broader pediatric, literature, we aimed to explore different stakeholders' views and recommendations with implementing education of patients about their DSD. We explored individual, parent, provider and allied professional perspectives of *who* should be involved in the process, *what* information should be shared and *when*, *how* the diagnosis and treatment should be discussed, and *why* educating patients about their condition is important.

2. Methods

The methods of the first phase of the *Defining Successful Outcomes and Trade-offs* (DSOT) study, a multicenter qualitative phenomenological study, have been described elsewhere.[30] Each of the three sites were children's hospitals and members of the US-based Differences of Sex Development – Translational Research Network.[31, 32] A summary and additional details pertaining to specific aspects of this project are provided below.

2.1 Participants

In this phenomenological qualitative study, trained interviewers with expertise in DSD (EW, KSJ, MG, TSK, DES) led individual or homogeneous, small-group interviews (based on participant preference), either in-person in clinic/research space or over the phone with teenagers and adults with a DSD (n = 24), parents of a child with a DSD (n = 19), healthcare providers specialized in DSD care (n = 37), and allied professionals with some familiarity/expertise in DSD (n = 30; e.g., chaplains, healthcare administrators, lawyers, medical ethicists, DSD researchers, social scientists, and support and advocacy organization leaders) (Table 1 and Figure 1). Eligible teenagers/adults with DSD (ages 15–40 years old) and parents of a child with a DSD (newborn to 15 years old) required a DSD diagnosis,[14] but those with Klinefelter or Turner syndrome were excluded unless urogenital atypicality was also present. Eligible healthcare providers were those from a range of pediatric specialties (e.g., child life, endocrinology, genetics, gynecology, neonatology, nursing, primary care and adolescent medicine, psychology, surgery, urology) serving patients with DSD. Allied professional (e.g., non-physician DSD clinical researchers,

healthcare administrators, lawyers, medical ethicists, support and advocacy organization leaders, and social scientists) eligibility included those involved in research, advocacy, or other professional work within DSD. Participants were recruited using stratified, random sampling across stakeholder groups as previously described.[30] Peer nomination/snowball sampling was also utilized for providers and allied professionals. Recruitment of teenagers/adults with DSD and parents occurred in person or via email/letter and then phone call at three pediatric medical centers in the US and Accord Alliance.[33] Healthcare providers and allied professionals were contacted via email for participation. Recruitment continued until thematic saturation was achieved across all groups. Additionally, two to three participants were recruited per specialty/profession across provider (e.g., gynecology, nursing) and allied professional groups (e.g., lawyer, social scientist). Each site's Institutional Review Board (IRB) formally ceded oversight to the lead site's IRB where ethical approval was granted. Consent was obtained from the parent and/or adult with DSD; assent was obtained from the teenagers.

2.2 Procedures

Interviewers used a semi-structured interview guide (Appendix A) and audio-recorded sessions, which lasted approximately 60 minutes. During interviews, only participant(s) and the interviewer were present. Overarching project aims included identifying successful patient outcomes across the developmental trajectory to integrate these considerations into clinical practice to improve DSD healthcare delivery. In addition to the primary questions of interest ("What is a successful outcome?" and "How do we achieve it?"), interviewers prompted participants to discuss topics specific to patient education about their condition and its implications (e.g., "Who first told you about your condition?" and "What did they say?"; "What are some of the things you have told your [patients/child] about [his/her] condition?" and "How old were they?" Participants also provided sociodemographic information.

2.2 Data Management and Analysis

Interviews were audio-recorded, transcribed, and coded (See Suorsa-Johnson et al., 2021 for coding details). A codebook was developed by the research team to identify themes, based on study aims, the interview guide, and preliminary themes that emerged from interviews. The codebook was modified to accommodate additional themes identified through coding. Utilizing NVivo 12 (QSR International, Victoria, Australia), co-authors (KSJ and AB) completed paragraph-level transcript coding. Twenty-five percent of transcripts were double coded for inter-rater reliability (92% agreement) at the beginning of coding and intermittently thereafter. Disagreements were discussed and resolved. To examine stakeholder perspectives related to patient education, eight codes related to education and communication with patients were examined. These included codes for: 1) patient autonomy, 2) patient self-efficacy, 3) patient knowledge and understanding of the condition, 4) teamwork/multidisciplinary team care, 5) multidisciplinary teams utilizing communication, honesty, and openness, 6) patient and family having a good understanding of intervention options/plan of care (e.g., informed consent), 7) patient and family educated about intervention options/approach to decision making/condition in general, and 8) patient and family are involved in decision making. To ensure quotes were not missed, additional

text searches of all transcripts were completed for words/phrases related to 1) providing developmentally appropriate information: “developmentally appropriate, age-appropriate”; 2) education: “educate, teach, language”; 3) language related to withholding or sharing information: “secret, disclosure, withhold, truth, share” and; 4) needing to educate more than once: “repeatedly, over time, all at once, segmented”. Transcript excerpts for relevant codes and text searches were reviewed by 3 investigators (EMW, KSJ, and AB). Authors independently reviewed themes and collaboratively organized and consolidated themes to ensure reliable analysis. Relevant themes were identified and organized according to “Who?”, “What?” “When?”, “How?”, and “Why?”. Educational themes were reviewed by the larger research group and modified following group discussion. Illustrative quotes are described in the Results (Table 2).

3. Results

Participants provided varied perspectives for *who* should educate, *what* information to provide and *when*, and *how* information should be given. However, differences were specific to each individual’s experience and not necessarily related to their stakeholder status. There was general agreement about the importance of educating children and teenagers about their DSD and DSD-related medical care (*why*).

3.1 Who?

Participants provided a range of opinions on who should educate patients. Some thought this task was best approached jointly by parents and healthcare providers. Others recommended parents should undertake conversations to educate their child because they were in the best position to offer emotional support. When considering the role of specialists, it was noted that “*team-based education is really important*” (social scientist) and most participants thought that having a mental health provider help facilitate education and provide psychological support was beneficial.

Although parents were suggested as the primary educators, some providers reported that parents may resist having open discussions with their child. Reasons included wanting to protect their child, uncertainty about what to say due to lack of their own understanding of the condition, and discomfort with discussions around sex and genitals. For patient education to occur, “*parents and caregivers […] have to be able to face their own shame, their own fear of marginalization, and their own fear of uncertainties around [sharing the condition with their child]”* (primary care provider). Furthermore, some individuals with DSD felt that parents should not have a choice to withhold information from their child. As an adult with DSD noted: *“parents shouldn’t have a choice, at all, in disclosing or not disclosing.”*

3.2 What?

Individuals with DSD and some parents agreed that sharing *all* the information was beneficial, and that honesty about the medical care patients had received was preferred. Specifically, participants expressed that “*honesty is the best policy*” (parent), and “*no matter if [the information provided] is good [or] bad*” (teenager with DSD). However, some felt too

much information could be overwhelming, suggesting the need to strike a balance between providing too much versus just enough. Despite the general suggestion, a specific approach to providing just the right amount of information was not identified.

Honesty and openness extended to providers being forthcoming about what they knew and what they did not know: "*In thinking about getting to a positive outcome […] the first piece is to be really open with families and youth about what we know and what we don't know. What are the limits based on research findings?"* (psychologist).

Individuals with DSD, providers and allied professionals felt it was critical to share medically relevant information. Furthermore, the patient should know what procedures they had undergone. Participants also provided specific recommendations of what information to share, including explaining the biological basis for the condition, the resulting phenotype or symptoms, discussing variations in development to help normalize the condition/physical difference, talking about the distinctions between privacy and secrecy, and reassuring the patient that there are others with similar conditions and experiences.

3.3 When?

Participants thought it was important to educate patients using a graduated and iterative approach. However, interviews lacked suggestions regarding how and when specific information should be introduced or over what period of time. A nurse noted this process would involve providing the child with *"years and years of information"* in *"slow little bits of information at each visit"*. Although recommended, this did not always happen in practice. As one patient noted: "*That's one of the things that I wish my parents could take back is the fact that they just told me all in one, like, spoonful. […] It definitely was [overwhelming]*" (teenager with DSD). A gynecologist believed that parents felt overwhelmed not knowing when to share information: *"The problem that's so scary for parents is they feel like they have to share everything right away. And I really think it's a process. And you share it in steps that are age-appropriate*."

One approach was to allow the child's curiosity to prompt information-sharing. As a chaplain stated: "*Children are going to let the parents know. Because the children are either going to have questions or have behaviors that are going to demonstrate that they want to know.*" Parents noted letting their child know they are available to answer questions and others stated their child had already asked about a range of topics (e.g., about body differences, medications, etc.). One barrier to waiting for children to bring up questions to parents or providers was that they might not feel comfortable: *"I didn't feel comfortable enough to ask the teacher about [genital differences], especially not in the class. And I didn't feel comfortable with asking my parents about it 'cause I just thought that either they knew or they didn't*" (teenager with DSD).

Looking to the medical specialist to identify when to share information was another approach. However, there was a lack of insight as to how providers would operationalize identifying when to share information. Many referenced providing "*developmentally appropriate*" information. When asked to identify a specific age when children should be informed about their diagnosis, participant recommendations ranged from toddlerhood to

the teenage years. Other participants thought it was difficult to identify a specific age, as each patient and family is different. However, "puberty" was generally identified as an appropriate time to move forward with education.

3.4 How?

Participants mentioned the language and tone used in imparting information. Recommendations included using simple words, avoiding pathologizing language, and adopting a neutral or positive tone. Suggestions were also made to frame discussions to exclude the word "normal." Having patients talk to others who were older and had lived with DSD was viewed as beneficial.

The necessity of repeating the same information over and over was noted by participants, as *"different things become salient, at different ages. […] They may not understand everything that is said to them when they are younger in the same way they will understand it when they are older"* (psychologist). One adult with DSD recalled remembering parents and providers *"explaining it to me and not understanding"*. However, individuals with a DSD who noted confusion when they were young children reported this resolved over time as information was re-explained as they became older. The need to repeat/clarify information could be identified by assessing patient understanding.

Participants stated the benefit of providing informational materials to parents and patients, such as booklets or online resources. However, others noted that accurate and helpful information was hard to come by. Participants also suggested providers should develop a strategy to help parents identify education topics and practice sharing information about the DSD condition with their child. Another family noted taking photographs of their child's anatomy before each surgery "*so he could see like what he looked like beforehand and […] what decisions we made for him*" (parent).

Other strategies recommended by participants included taking advantage of or even creating opportunities to share information. This suggestion often arose when participants discussed having to educate individuals about their infertility. For example, when daughters pretended to be expecting a baby, parents would capitalize on their play behavior to teach their child about alternative ways to build a family, openly discussing adoption options with their child. One parent chose to use the adoption of a pet to discuss the potential role of adoption in their child's future. Providers also suggested how parents can utilize opportunities to educate; for example, discussing and normalizing adoption or alternative ways to have a family.

Another facilitator to help families was the ongoing conversation between providers and parents regarding the education process. Providers should assess parents' own understanding, if parents have talked to their children, what they have discussed, and how the conversation went. *"I think health literacy in this population would be really important to assess. And then kind of individualize your care of the family based on, you know, how much they understand, how much they want to understand, and their relationship with their child"* (clinical researcher).

3.5 Why?

A successful outcome noted by participants was one in which the patient had a general understanding of what was happening with their body and the ability to make informed decisions about their care as they were old enough. Individuals with DSD stated that, once they were aware of their condition and the medical care they had received, the benefits of education were apparent. For example, the condition and life in general became easier to manage and a full understanding helped to promote positive adjustment.

Furthermore, if patients were not educated about their condition, participants worried that patients would turn to unreliable sources of information, such as asking friends, searching the Internet or referencing pornography. As a gynecologist stated: *"People are much more aware of their genitals than they were ever in the past because of the internet and because of porn."* In addition, distrust, shame and secrecy could occur if patients did not receive accurate and honest information about their condition: "*you don't want that trust bond [between parent and child] to be broken by [parents] withholding information long-term*" (nurse).

Finally, managing both parent and patient expectations was a common reason cited for educating patients. All participants noted that patients developing clear expectations regarding their DSD and the processes related to their conditions could help determine better long-term outcomes. A father reported that *"it helped [our daughter] with just understanding, 'well, okay, I need to start watching out for this' or 'this is what I can expect in the next year or whenever'… It put her mind at ease and our minds at ease, I think."*

4. Discussion and Conclusion

4.1 Discussion

We sought to explore how a range of DSD stakeholders valued patient education and their perspectives on this topic. This study demonstrates the complexities involved when considering how to share information about a DSD with children. It was clear that educating patients about their condition and practicing openness during those conversations are crucial. Participants' insistence on education across stakeholder groups is consistent with DSD practice guidelines and the broader pediatric literature.[1, 4–6, 14, 34] Participant suggestions align with recommendations about educating children with a DSD and their parents about the condition from a pediatric psychologist and suggestions from adolescents and young adults with DSD about infertility education.[35, 36]

Some parents felt that thoroughly educating the child about their DSD was the "best policy". However, some parents could be reluctant to share information with their child, which could pose a barrier; educating parents on how to in turn have these conversations with their child could facilitate the process. Among girls with congenital adrenal hyperplasia, parents have reported concern that if their children learned about their genital surgery, it would lead to changed self-perception, an altered perception of parents, and confusion.[37] There are reports of children initially reacting negatively with maladaptive behaviors, but that these negative reactions and behaviors decreased over time and, ultimately, education resulted

in improved psychosocial outcomes.[38] Other research suggests that education about the medical condition is not what leads to child maladjustment, but rather the parenting role or style may be what results in negative reactions.[39] Future research should address these concerns regarding potential maladjustment or negative perceptions when educating children about their DSD condition.

Although some participants offered general ideas and principles on how to go about educating patients, instructions to guide parents or providers on how to have education-focused conversations were vague. The extant literature is consistent in recommending provision of “developmentally-appropriate” information to children and adolescents, but with few specifics on implementing this approach across age groups or developmental stages. Even the 2006 Consensus Statement and 2016 Update, while attempting to delineate best practices and recommendations on educating patients about their condition, offer no specific guidance. Consistent with the broader DSD literature, the lack of detailed guidance from our participants is a limitation of our study and highlights an area of need. Fortunately, there are some published suggestions on how to operationalize education for children with other medical conditions, such as educating children regarding their HIV status or their parent’s HIV status, that could be extrapolated for those with DSD.[40–44] While literature on educational interventions is lacking in DSD, Slijper and colleagues provided suggestions on the process of educating individuals with androgen insensitivity syndrome (AIS).[20] This included parents educating the child, first, about typical sexual development, then by providing all information about AIS except the XY chromosomes at age 11, and finally sharing about the sex chromosomes around age 16 or 17. However, research has not tested the utility of this recommendation and, though somewhat generalizable, it is limited to one condition. Therefore, more research is needed in this area to identify effective approaches for providing information across the developmental spectrum.

Differing opinions on when to provide education is similar to other studies. Parent preferences for age of informing their daughters with congenital adrenal hyperplasia about their early genital surgery ranged from younger than 9 to over 18 years.[37] Participants generally regarded “puberty” as the best time to have a broader education session. Puberty can be highly variable with each diagnosis that falls under the DSD umbrella. This also does not address best practices surrounding pre-pubertal children and what they should be told and when. There were also differences of opinion over whether education should be undertaken by providers versus parents with assistance from providers. This lack of clarity in who should educate the child also exists in other pediatric conditions, including genetic conditions.[45] Blankstein and colleagues discovered that although almost all parents of boys with hypospadias planned to educate their child about their hypospadias repair, 90% denied receipt of information from providers about how and when to do this.[46] This suggests a lack of meaningful integration of recommendations into practices that support patient education.

Whether providers are educating patients regarding their condition or educating parents about how to communicate information to their child, there is a need for providers to ensure they are using lay-person language and even-tone in discussions as well as ensuring they have a strategy to help facilitate this process. Providers should also be aware of

perceived or actual stigma experienced by the patient or family, as these could negatively impact the patient.[47] Patient education should be supported by evidence-based educational interventions and include resources for parents and children/teenagers with DSD.

Participants stated that resources such as those available through websites or books may be beneficial, although some thought that finding accurate resources was a barrier. The struggle to identify reliable information, particularly online, is not unique to DSD.[48] Fortunately, a vetted DSD-specific online resource repository exists to which families can be directed.[49] Different stakeholder perspectives emphasize that there is more than one way to educate children with DSD about their condition, underscoring the importance of individualizing care.

4.2 Conclusion

Educating children about their DSD is a key element of patient-centered care. The qualitative interviews from our study provided important insight from teenagers and adults with DSD, parents of children with a DSD, healthcare providers, and allied professionals about the who, what, when, how, and why of educating (Table 2). Providers play a key role in supporting patient education. There are barriers to education and providers need to be aware of these and address this gap in care. Clarification is needed to identify specific strategies, especially regarding details of providing developmentally appropriate information to patients.

4.3 Practice implications

Healthcare providers are essential in ensuring patient education about DSD, both by delivery of information directly to their patients and by educating parents on how to share information with their child. Within this process of supporting education, providers should assess for, and aim to, understand the underlying reasons for parental resistance with respect to patient education, whether it be cultural, stigma, lack of knowledge or fear of the unknown. Working through these barriers, providers and parents can have meaningful conversations which are essential to ensure successful patient education which is the overarching goal in providing complete DSD care. Although the specific approach to patient education may look different across families depending on cultural or familial dynamics, supporting education should not be sacrificed due to parental resistance.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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Highlights

- Who: Providers and parents play a key role in supporting patient DSD education.
- When: Educate gradually, considering patient questions and provider input.
- What: Strike a balance between providing too much versus just enough information.
- How: Use simple, positive language, educational materials, and repeat information.
- Why: Educating children about their DSD is a key element of patient-centered care.

Practice implications:

Healthcare providers are responsible for supporting the education of children and teenagers with DSD about their condition. When considering barriers, adopting a cultural or family systems framework can reduce parental resistance and promote open dialogue.

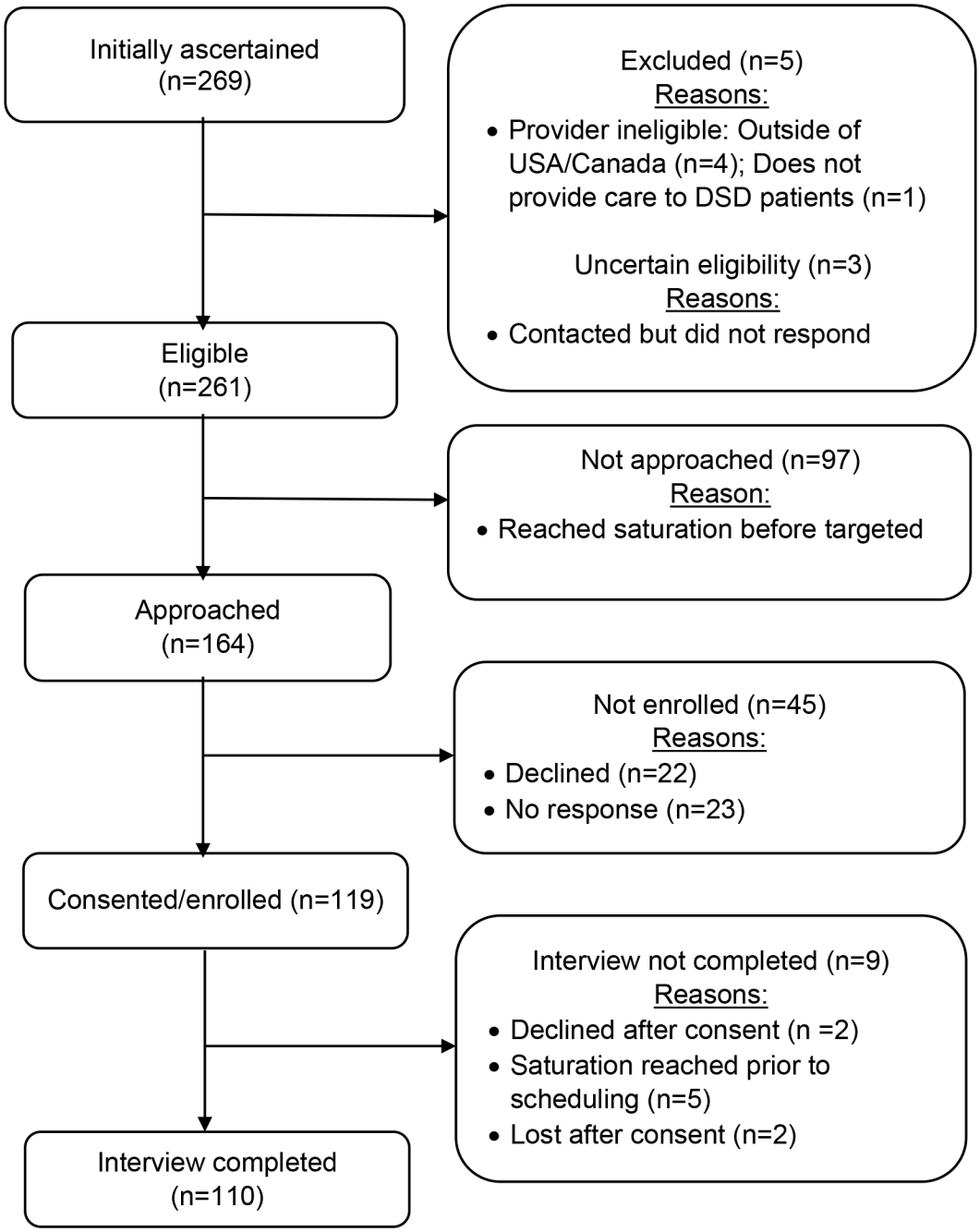

Figure 1.
Recruitment Flowchart

Table 1.

Participant Demographics

	Individuals with a DSD & Family Members		Healthcare Providers (n = 37)	Allied Professionals (n = 30)
	Individuals (n = 24)	Parents (n = 19)		
Age, years; Mean (SD), range	22.1 (7.1), 15–39	37.8 (6.9), 25–52	46.1 (9.4), 28–68	49.9 (12.2), 32–78
Gender identity, n (%)				
Boy / Man	1 (4.2)	6 (31.6)	7 (18.9)	14 (46.7)
Girl / Woman	22 (91.7)	13 (68.4)	29 (78.4)	15 (50.0)
Other	1 (4.2)	0	1 (2.7)	1 (3.3)
Race, n (%)[a]				
African American / Black	3 (13.0)	1 (5.3)	1 (2.7)	1 (3.7)
White	16 (70.0)	17 (89.5)	30 (81.1)	24 (88.9)
Other / more than one race[c]	8 (17.4)	2 (5.3)	6 (16.2)	3 (10.7)
Hispanic	5 (20.8)	3 (15.8)	1 (2.7)	0[b]
Sexual orientation, n (%)[a]				
Straight/Heterosexual	14 (63.6)	15 (93.8)	34 (91.9)	22 (84.6)
Other[d]	8 (36.4)	1 (6.3)	3 (8.1)	4 (14.3)
DSD diagnosis / category[e,f]				
5α-reductase deficiency	0	1 (5.3)		
17β-hydroxysteroid dehydrogenase deficiency	0	1 (5.3)		
46,XY DSD (not otherwise specified)	2 (8.3)	0		
Ambiguous genitalia	0	2 (10.5)		
Androgen insensitivity				
Complete androgen insensitivity syndrome	4 (16.7)	0		
Partial androgen insensitivity syndrome	2 (8.3)	1 (5.3)		
Cloaca / cloacal exstrophy	3 (12.5)	4(21.1)		
Complete gonadal dysgenesis	2 (8.3)	0		
Congenital adrenal hyperplasia	7 (29.2)	3 (15.8)		
Hypospadias	0	3 (15.8)		
Mixed gonadal dysgenesis	2 (8.3)	0		
MRKH syndrome / Mullerian anomaly	2 (8.3)	1 (5.3)		
Ovotesticular DSD	0	2 (10.5)		
Healthcare Provider Specialties				
Endocrinology			6 (16.2)	
Genetics, Genetic Counseling, Genomics			6 (16.2)	
Pediatric & Adolescent Gynecology			4 (10.8)	
Primary Care & Adolescent Medicine			6 (16.2)	

	Individuals with a DSD & Family Members		Healthcare Providers (n = 37)	Allied Professionals (n = 30)
	Individuals (n = 24)	Parents (n = 19)		
Patient Education & Counseling				26
Psychology			5 (13.5)	
Pediatric Urology & General			4 (10.8)	
Surgery				
Other: Child Life, Neonatology,			6 (16.2)	
Nursing				
Other Professions				
Chaplaincy / Pastoral Care				7 (23.3)
Healthcare Administration				4 (13.3)
Law				2 (6.7)
Medical Ethics				4 (13.3)
Research, Clinical				5 (16.7)
Research, Social Science				4 (13.3)
Support & Advocacy Organization				4 (13.3)
Leadership				

Note.

[a]Percentages adjusted for missing data;

[b]1 participant declined to answer and 1 missing;

[c]Other = Asian, Hawaii Native / Pacific Islander, or Other;

[d]Other = Lesbian, gay, homosexual, or bisexual;

[e]For those recruited through clinic, diagnoses were derived from chart review; for those recruited through support or advocacy organizations, diagnoses were self-reported (n = 8);

[f]For parent participants, “diagnoses” reflect child’s DSD condition;

SD = standard deviation; DSD = difference of sex development; MRKH = Mayer-Rokitansky-Küster-Hauser syndrome

Table 2.

Exemplar Quotes

Themes & Subthemes	Exemplar Quotes
Who?	
Parents versus providers	"Parents should definitely be able to tell their kids that and help them and hold their hand through that because if you do love your parents, that's super-duper helpful. But I do think that healthcare providers can tell you more about [your condition] that your parents just can't understand 'cause not everybody is a medical professional. So it's good to have that side information. But for the main information, just the range of what's happening to you, I think that should come from the parents." - Teenager with DSD
Providers	"And if the parents don't feel comfortable, or not well equipped to find words to [educate the child], or for us to do it with the parents in the room if they prefer that, whatever works with that particular family." - Gynecologist
Parents	"I think it would be best coming from your parents just because, like, it's like an emotional thing to have said to you. So, if you have a provider, or like someone that you're not super... Sure if you're super close to that one person, then I would understand. But if you're not really close to the person, then you're just kinda stuck in this whole little ball of just emotions. And your parents can probably be there for you." - Teenager with DSD
Barrier: Parental comfort	"You know, there are some families where talking about sex is like no big deal we do it all the time, whereas there is other families where the word "sex" already makes everybody uncomfortable, well then you get a situation like this on top of that makes it really, really hard for some families." - Gynecologist
What?	
All the information versus some	"I am perfectly fine if I don't have every single detail, but as long as I've got more than just general information...like a little into detail, but not completely overwhelmed on every little single little thing" - Teenager with DSD
Accurate/honest information	"And I think being honest is the most important thing, that you aren't doing any favors to the patient or the parents by not being honest. And so that was a big issue throughout all of my care, was the lack of transparency, the lack of honesty, and really having to seek our own answers for stuff that had been done since birth, really. And that shouldn't be the case, if there were mistakes made, or just based on the disorder itself, if things aren't options or if things are options, then those need to be made clear." - Adult with DSD
Relevant information about medical condition and procedures	"I do think that in order to really have the autonomy, to have a voice, in making those decisions around some of those tradeoffs, they do have to have, you know, a good understanding of their medical condition and their history of any previous, you know, surgeries that they have had in, sort of, making some of determinations later on." - Psychologist
Topics covered	"We start with a general education about how bodies develop, initially, along the same lines and then how male and female development go separately and how there are lots of things that can happen during that process and then trying to move into their diagnosis, their presumed diagnosis, through that pathway." - Urologist Privacy versus secrecy: "Some of them are like 'oh yeah,' and the other ones, it doesn't make a difference. I try to do it lighthearted. I think some people appreciate that and can go 'yea, you're right, nobody needs to know.' [...] I'm sure because they had a surgery or because they. what they think is different - they're worried about it. But reminding them that: 'unless you share that with someone, a lot of people will never even know.'" - Urologist
When?	
At a developmentally appropriate age	"So I think it's going to have to be individualized, but I think keeping the child's best interest in mind so there is a medical or safety risk to the child, then you might need to discuss it on an appropriate developmental level with the child and perhaps with daycare or teachers when they are younger, but I think it varies. You can't totally obscure the reason that kids are going to the hospital or having blood drawn or having imaging tests done... Just like we would for cancer, for diabetes, for asthma, we kind of talk about things at a level the child can understand and allow them to ask questions and encourage them to keep asking questions." - Primary Care Provider
Early enough to manage expectations	"We moved here when she was in 5th grade. So she hadn't started her period yet. So we got to see [a surgeon] and be her patient for a year or so, before." "Before [the surgeon] did anything. But it helped [daughter] with just understanding, 'well, okay, I need to start watching out for this' or 'this is what I can expect in the next year or whenever.' That's helpful. It put [our daughter's] mind at ease and our minds at ease." - Parents
How?	

Themes & Subthemes	Exemplar Quotes
Simple words, avoid pathologizing language, adopting a neutral/positive tone	"We consciously avoid pathologizing language, even with her adrenal insufficiency. [...] It most certainly is a medical disorder, but we don't want her growing up feeling that she is "diseased" or that there is something "wrong" with her, and so we call her cortisol her "cortisol," not "medicine." Medicine is for when you're sick. And your cortisol is something that you don't make. Some bodies make it, some bodies don't. Her body is one that doesn't." - Parent "Being pretty straight forward with kids about it and if their anatomy looks different than their siblings, to have a simple explanation for why that is and not to belabor it. [...] definitely not having it in a shaming way, having it in a way that: 'people have different colored hair, [...] people are tall or short, some people are fatter or thinner, people have different genital appearance.'" - Primary Care Provider "I often talk to parents like that if they cry and when they are talking to kids about their conditions and how sad they are and how terrible it is, I think kids will internalize their parent's perspectives and emotional responses and that might instill fear and a feeling of unhappiness. So I think the way parents present information in their own emotional presentation and nonverbal presentation is all very important." - Psychologist
Assess understanding	"I am looking for whether they know their diagnosis. What do they understand about how their development progressed a little differently than other kids? What do they know or what have they been told about any sort of past medical history? What do they understand about how their medical condition may impact them in the short and long term?" - Psychologist
Provide resources and be supportive	"I still think it's good to watch videos about people sharing their story, 'cause it shows what they did, what's happening with them now. And I think it's. It helps more, you understand more, and it helps you think about all of your options and what you think works for you." - Adult with DSD "What we do in our teen-based clinics, we really have created all visits have an element of empowering and safe-space. Where, as kids grow, one of the first questions that's asked at every visit, every time is "how are you feeling? What's new with you? And what can we do to make your life better or what questions do you have today about you?" And letting the youth lead it. So parents see from our actions that we're about empowering kids." - Primary Care Provider
Utilize teaching moments and create opportunities	"Let's say you meet a family who has adopted a child, just say, 'oh yeah, some families adopt and some families have a baby on their own or some people have a baby where somewhere else carries the pregnancy.' [...] I encourage families to start bringing that up early [...] if infertility is going to be a reality for their child." - Gynecologist
Normalize	"I think that goes back to education. I think we need lots more education about variations in development, reproductive anatomy. We spend a lot of time going over that and how varied everyone is...I feel like patients should have a very clear understanding of their condition and understanding of how there's a natural variation in human development instead of 'disorder' or something that's 'abnormal' -- especially when they're trying to adjust." - Gynecologist
Why?	
Easier to manage condition, empowerment, acceptance	"So it became easier once I knew. For sure, it became easier for school, I mean, I just stayed focused, but knowing something was off, it was a lot to going on...But once I found that out, it was a lot smoother....And it just got a lot easier dealing with it once you knew. Once you are aware because you feel like a crazy person because you didn't know what was going on." - Adult with DSD
Promote positive adjustment	"Education about medical history can impact long-term adjustment: "how open [parents] are with their kids, you know, about decisions that maybe they made about not doing early surgery -- I think that can also play into adjustment long-term." - Psychologist