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Future fertility for individuals with differences of sex development: Parent attitudes and perspectives about decision-making

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Summary

Background
Children, adolescents, and young adults (children/youth) with differences/disorders of sex development (DSD) face challenges related to future fertility; this may be due to variations in gonadal development, and, for some, gonadectomy performed to reduce the risk of malignancy. Childhood may be the only time for preservation of biological fertility potential for children/youth who undergo gonadectomy or have early gonadal failure. Fertility-related decision-making for these patients is particularly complicated, due to the need for parental proxy decision-making, potential discordance between gender identity and gonadal type, and uncertain future assisted reproductive technologies.

Objective
This study aimed to assess: (1) attitudes regarding future fertility, and (2) healthcare needs for fertility-related decision-making among parents of children/youth with DSD.

Study design
Semi-structured qualitative interviews about future fertility were conducted with parents of children/youth with DSD. Parents who had never discussed fertility with a healthcare provider were excluded. Grounded theory methodology was used to identify emergent themes and patterns. Demographics and clinical characteristics were assessed via survey and medical chart review.

Results
Nineteen parents were interviewed (participation rate: 60%, 14 mothers/5 fathers, median patient age at diagnosis 6 months (range 0–192), eight DSD diagnoses). The most common emergent themes are summarized in the Summary Table. Most parents identified fertility as a key concern, both at time of diagnosis and throughout development. Parents expressed difficulty with timing of disclosure about potential infertility to their children. Multiple preferences related to medical decision-making about future fertility and fertility preservation were expressed, including: a desire for step-by-step decision-making, and use of medically vetted information and research to guide decisions.

Discussion
This qualitative study provided new information about the perspectives of parents of children/youth with DSD regarding future fertility. Previous studies have suggested that the possibility of biological parenthood is important to many individuals with DSD. This study provided an in-depth parental perspective. This is important because many decisions that affect future fertility are made in childhood, and require parents to make decisions on behalf of their children. The study sample was limited in its geographic diversity. Strengths of the study included diversity in age of the child/youth, ethnic backgrounds, and the DSD diagnoses that were represented.

Conclusions
Future fertility was a concern for many parents of children/youth with DSD. Parents expressed multiple priorities and preferences related to making difficult fertility-related medical decisions for their children. Many of the study findings could be incorporated into future best practices for discussions about fertility with families of children/youth with DSD.

Summary Table Parent attitudes toward fertility, disclosure, and decision-making: common themes.

Theme	Frequency
Topic 1: Attitudes toward fertility/infertility	
Fertility status of child/youth 'unknown'	12
Importance of child/youth being able to have their own child	12
Fertility as a main concern	10
Concern about child dealing with fertility status in the future	10
Topic 2: Attitudes toward disclosure	
Struggle with timing of diagnosis/fertility discussion with child	9
Openness with family/friends	8
Selective disclosure to family/friends	8
Topic 3: Decision-making about child's future fertility and fertility preservation	
Desire for research, medically vetted information and statistics to guide decisions about fertility preservation/gonadectomy	17
Parents desire to take things step by step	16
Desire to discuss child's condition with other families	16
Alternate ways to have a child	12

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Introduction

Disorders/differences of sex development (DSD) encompass a range of conditions in which chromosomal, gonadal, or anatomic sex development is atypical [1]. Many children, adolescents, and young adults (henceforth children/youth) with DSD face challenges related to future fertility; this may be due to: (1) variations in gonadal or reproductive tract development, (2) prophylactic gonadectomy performed for malignancy risk, (3) progressive gonadal failure, and (4) hormone replacement therapies [2–4]. Some children/youth with DSD may be eligible for fertility preservation (FP) options, including experimental cryopreservation of gonadal tissue. Others may not possess biological fertility potential within the limitations of current assisted reproductive technologies (ART). Childhood may be the only time for preservation of biological fertility potential for children/youth who undergo gonadectomy or have early gonadal failure.

Understanding healthcare and decision-making needs related to future fertility and FP for patients with DSD is complex. Each family's experience and preferences vary depending on factors such as the child/youth's age and diagnosis, cultural, religious, and family values. Compounding the complexity is the potential discordance between gender identity and gonadal type, uncertain future ART, and the unknown fertility potential of many DSD conditions. Additionally, decisions about treatments that affect future fertility and decisions about FP are often made by parents, rather than children/youth themselves.

Fertility potential has been reported for some DSD conditions, including: Klinefelter Syndrome (KS) [5–7], Turner Syndrome (TS) [8,9], congenial adrenal hyperplasia (CAH) [10], and ovotesticular DSD [11]. However, fertility potential has not been completely defined for many DSD conditions. Unknown fertility potential, which is associated with many DSD conditions, may lead to stress, confusion, and difficulty making treatment decisions that affect future fertility for individuals with DSD and their families. Studies of attitudes towards future fertility and FP in individuals with DSD suggest that future biological parenthood is a priority and concern [12,13]. However, such studies are limited by small sample sizes and non-systematic data collection.

Given the potential complexity of fertility-related decision-making in DSD, and the paucity of literature on this topic, this study sought to address this gap by assessing: (1) attitudes regarding future fertility, and (2) healthcare needs for fertility-related decision-making among parents of children/youth with DSD.

Material and methods

Study sample and recruitment

Parents of children/youth aged 0–30 years with a DSD diagnosis were approached between October 2015 and December 2016 by telephone or in person during clinic. Recruitment brochures were made available to potential participants at Lurie Children's Hospital and the four institutions of national colleagues. Non-English speaking parents, and parents who had not previously discussed their child's fertility with a healthcare provider were excluded (to avoid undue distress from first introducing fertility during a research study). Written consent was obtained before each interview.

Qualitative interviews

Two trained interviewers conducted semi-structured qualitative interviews. Parents were asked open-ended questions about: (1) medical care received for their child's DSD condition, and views on their child's ability to have a biological child; (2) disclosure of their child's fertility status to the child/others; and (3) decision-making regarding surgery and FP (Appendix 1). Parents also completed a survey regarding demographics and clinical information about their child's diagnosis (also confirmed by chart review) and gonadal status. Interviews were audio-recorded.

Qualitative analysis

Recordings were transcribed verbatim and de-identified. Grounded theory methodology using a constant comparative approach [14,15] was employed to develop a coding scheme within the framework of the three topic areas. Two researchers, who assigned codes to the text, independently read the initial transcripts. Coding rules were established by discussion and consensus. The remaining transcripts were coded using this scheme, and the codes were modified and expanded until saturation was achieved (no new themes emerged). Coding was facilitated by MAXQDA qualitative analysis software (Berlin, Germany). Frequencies of themes/subthemes and representative quotations were compiled.

Results

Study cohort

Of the 28 consecutive families who fitted inclusion criteria, the study team approached 22 families (32 parents), and all agreed to participate. Of these, 19 participated in an interview (18 from Lurie Children's Hospital and one from an outside clinic whose practitioner referred the parent to the study team; the participation rate was 60% of individuals initially approached, 14 mothers/5 fathers). One participating parent had two children with a DSD diagnosis, and parents from five families both participated (interviews completed separately). Demographics are detailed in Table 1. Median age of the child/youth at diagnosis was 6 months (range 0–192) and 7.5 years (range 0.5–22) at the time of parent interview. Eight DSD diagnoses were represented.

Thematic content

The following topic areas and corresponding emergent themes were presented in detail: (1) attitudes toward fertility/infertility, (2) disclosure/communication, and (3) fertility-related decision-making. Tables 2–4 outline frequencies of themes and representative quotations.