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# Enhancing intersex healthcare: A qualitative study of parental perspectives on the role of genetics

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## Abstract

Intersex individuals, encompassing people with diverse sex characteristics that do not fit binary frameworks of sex, have long faced a history of medical secrecy, discrimination, and societal stigma, contributing to their limited social visibility. In recent years, increased awareness of intersex issues and a robust advocacy movement have drawn significant attention to the experiences of intersex individuals and their families. This study contributes to the existing literature by examining the experiences and needs of parents of intersex individuals within genetic healthcare systems, bridging a critical gap, and advocating for more comprehensive and supportive healthcare practices. Semi-structured interviews were conducted with 14 parents of intersex individuals, and reflexive thematic analysis was used to inductively generate four major themes. Themes highlighted the need for improved accessibility of intersex healthcare, the importance of multidisciplinary healthcare teams, and the significance of clinical diagnosis provided by genetics professionals. Furthermore, the study highlighted the necessity of a thoughtful approach to information provision and the impact of genetic investigations on family dynamics. Genetics professionals can play a pivotal role in raising awareness about intersex variations, improving diagnostic processes, collaborating within healthcare teams, and providing specialized support to address psychosocial concerns. The study underscores the importance of treating families as a collective entity and addressing the impact of genetic investigations on the family unit. By addressing the challenges and implementing the recommendations outlined, healthcare institutions can create a more compassionate, inclusive, and effective healthcare environment for the intersex community.

**KEYWORDS**
caregivers, differences of sex development, family, genetics services, intersex, variations of sex characteristics

Kayla Horowitz and Kimberly Zayhowski contributed equally and should be considered joint first authors

## 1 | INTRODUCTION

Intersex individuals have diverse sex traits that challenge binary definitions of male and female bodies, encompassing a spectrum of variations in gonads, genitals, secondary sex characteristics, hormones, and chromosomes (Office of the High Commissioner for Human Rights (OHCHR), 2015). Diversities within the intersex community question established societal norms related to sex and gender. Despite constituting an estimated 2% of the population, intersex individuals have often remained hidden from public awareness due to the prevalence of medical secrecy, discrimination, and societal stigma (Crocetti et al., 2020; Haghighat et al., 2023). The language surrounding intersex, variations of sex characteristics (VSC), and disorders/differences of sex development (DSD) has evolved over time, reflecting changing perspectives and priorities (Viloria & Nieto, 2020). In this article, we predominantly use the terms "intersex" and "variations of sex characteristics (VSC)" for several reasons, with the acknowledgement that some families may prefer other terminology. "Intersex" is a term embraced by some individuals within the intersex community as it can empower them to assert their identities and experiences. "VSC" is a more neutral umbrella term that refrains from pathologizing bodily diversity while ensuring inclusivity for individuals with VSCs who may not hold intersex as an identity (interACT and Lambda Legal, 2018). While "DSD" has been used in clinical contexts, it is critiqued for framing variations as medical disorders and reinforcing a sense of "otherness." By using "intersex" and "VSC," we aim to prioritize the agency, autonomy, and dignity of intersex individuals and their families, acknowledging that the language we employ can shape perceptions, attitudes, and the quality of care they receive.

Historically, the management of VSCs has followed a paradigm aimed at normalizing intersex variations through gender assignment and genital surgeries (Carpenter, 2018; Prandelli & Testoni, 2020; Roen, 2019). These interventions, often performed before the affected individual can meaningfully participate in decision-making, place the responsibility on parents to provide consent for their children. In the face of an unknown situation and a pathologizing medical discourse, parents often prematurely consent to these surgeries without full knowledge of their potential negative consequences or the realization that non-intervention is also a valid option, limiting their ability to make fully informed decisions (Carpenter, 2018; Liao et al., 2015; Sandberg et al., 2017). This surgical approach was founded on the unsupported belief that gender identity could be shaped and altered, and that modifying anatomical variations to conform to societal gender norms would alleviate psychological distress (Carpenter, 2018). However, these surgeries can exacerbate stigma and distress (Haghighat et al., 2023) and can lead to physical and psychological repercussions such as incontinence, chronic pain, mental health issues, and sexual difficulties that often persist into adulthood (Roen, 2019). The medicalization of VSCs has been further shaped by underlying homophobic, transphobic, and ableist ideals in physicians' attempts to diagnose one's "true" sex and resolve "physical ambiguity" in order to prevent "abnormal sexual behavior" (Crissman et al., 2011; Crocetti et al., 2020; Prandelli & Testoni, 2020). Societal norms, along with a lack of understanding of sex and gender diversity, create significant hurdles for intersex individuals and their families when seeking support (Haghighat et al., 2023; Lundberg et al., 2021).

The late 20th century marked the rise of intersex activism, sparking a pivotal shift in the biomedical approach to intersex care to focus on prioritizing individual autonomy (Carpenter, 2018; interACT and Lambda Legal, 2018; Lee et al., 2016). In alignment with this paradigm shift, the framework guiding this research is grounded in an agency-based model of intersex health (Crocetti et al., 2020). With the social model as its foundation, this model posits that centering intersex people's bodily autonomy is necessary in order to address the health issues faced by some people with VSCs as well as the sociocultural constructions that guide their medical treatment (Crocetti et al., 2020). The social model of intersex health is inspired by critical disability theory, which challenges the biomedical pathologization of intersexed embodiment and recognizes the disabling of intersex bodies as being a by-product of societal norms, prejudice, and discrimination (Crocetti et al., 2020; Koyama, 2003). An agency-based model of care places the individual with a VSC at the center of the decision-making process, and may reflexively support parents as they navigate their wider sociocultural context and bridge gaps in their medical care and service provision.

Genetic counselors possess a unique set of skills, a humanistic orientation, and a distinct role within the medical team that enables them to provide person-centered psychosocial education and support, and there is a call for inclusion of genetics providers

### What is known about the topic

Existing literature has offered insights into challenges faced by intersex individuals, stemming from issues like medical secrecy, discrimination, and societal stigma, as well as the detrimental impact of non-consensual surgical procedures. However, literature has offered limited insights into the experiences and challenges faced by parents of intersex children as they navigate the healthcare system.

### What this paper adds to the topic

This paper offers insight into the experiences of parents of intersex children in the healthcare system, emphasizing the need for enhanced accessibility, holistic multidisciplinary communication, and the crucial role of genetics professionals in intersex healthcare. Furthermore, the study underscores the importance of approaching families as a collective entity and recognizing the far-reaching impact of genetic testing on family dynamics.

on the multidisciplinary teams (MDTs) for intersex people (Gramc et al., 2021; Roen, 2019; Saulnier et al., 2021). Genetic factors contribute significantly to the development of intersex traits, thereby making genetics professionals essential to promote an understanding of these variations as well as any necessary treatment based on medical conditions that can co-occur with some intersex variations (Gramc et al., 2021; Haghighat et al., 2023; Martinez de LaPiscina & Flück, 2021). In parallel to the rare disease literature, individuals with VSCs and their families often have a long quest for a diagnosis or an understanding of the variation (Belzer et al., 2022; Simpson et al., 2018). Further, due to the legitimacy and acknowledgement that a medical label can provide, genetic diagnoses may grant access to resources such as appropriate medical intervention, informed decision-making, participation in clinical trials, and community engagement (Belzer et al., 2022; Jones, 2022). While the benefits of diagnostic clarity have been outlined in both rare disease and intersex scholarship, individuals receiving a diagnosis of a VSC may learn this information in a traumatizing or distressing manner, which can result in incomplete comprehension, challenges adapting to this information, and compounded stigmatization (Crocetti et al., 2020; Roen, 2019).

Genetic counselors' sensitivity and expertise in genetics can provide clarity regarding the genetic implications of intersex variations and facilitate informed decision-making. While genetics professionals can play a crucial role in supporting intersex individuals and their families, it is essential to acknowledge that historical misuses of genetic knowledge, exemplified by the eugenics movement, have contributed to the pathologization and discrimination of the intersex community (Berro & Zayhowski, 2023). In recognition of this history, medical documentation and diagnostic labeling must be thoughtfully considered, aligning with the individual's preferred language, avoiding pathologization, and incorporating person-first language. A nuanced and ethical approach to genetic counseling is paramount in promoting understanding, dismantling harmful stereotypes associated with intersex variations, and fostering acceptance, resilience, and well-being for intersex individuals and their families.

Delivering patient-centered and empathetic education about the diagnosis, treatments, and support provided by the MDT is crucial for intersex care (Saulnier et al., 2021; Weidler & Peterson, 2019). This approach enhances families' coping strategies, ultimately leading to positive outcomes for individuals with intersex traits, such as improved social acceptance and overall well-being. The existing literature underscores the profound impact of parents on their child's social and emotional development, as well as their capacity to cope with and adapt to their intersex variation (Sandberg et al., 2017; Wisniewski, 2017; Wolfe-Christensen et al., 2014). Additionally, evidence demonstrates that parents play a significant role in shaping how their child navigates their intersex identity and experiences (Schweizer et al., 2017).

The experiences of intersex people and their families in genetic care have been underrepresented in the existing literature. By placing intersex families' priorities at the forefront of healthcare practice considerations, genetics providers can reduce pathologization, minimize stigmatization, and optimize the coping mechanisms of both parents and children (Saulnier et al., 2021). This focus on parental experiences is essential, as it not only acknowledges their critical role as advocates for their children but also recognizes the profound impact their experiences can have on the overall well-being of intersex individuals. This study aimed to understand the needs of parents when navigating the healthcare system related to their child's intersex variation, as it is a crucial insight to advance VSC healthcare research and promote supportive healthcare practices.

## 2 | METHODS

### 2.1 | Study design

This research used a qualitative study design to explore the perspectives of parents of intersex individuals regarding their experiences and needs in healthcare settings, and genetics clinics specifically. Semi-structured interviews were conducted via Zoom videoconferences with parents of intersex individuals. Ethical approval for the research was granted by the McGill University Health Centre (MUHC) Research Ethics Board [2022-7864] in November 2021.

The study implemented an integrated knowledge transfer design, involving consultation with various key informants during its development to ensure the study met the needs of the community (Straus et al., 2013). These informants included representatives from McGill's DSD Group, prenatal and pediatric genetic counselors, a medical liaison from interACT Advocates for Intersex Youth, an intersex community member, and a parent of an intersex child, thereby fostering a comprehensive and community-oriented approach to the research. The author team comprised of genetic counselors, genetic counseling students, and bioethics research specialists. Additionally, many of the authors possess qualitative research and intersex health expertise. Several of the authors are queer, and none of the authors have known intersex variations.

### 2.2 | Participant recruitment

The eligibility criteria for this study included parents of children diagnosed with VSCs, who had access to a phone or the internet, and were English-speaking. Parents who had become aware of their child's VSC within the last year were excluded due to the sensitive nature of the topics discussed. Recruitment of parents of children with VSCs was facilitated through emails to various online support networks. Participants were asked to reach out to contacts who might be interested. Given the study's specific focus, defined sample population, applied theoretical frameworks, and high-quality in-depth interview data, data collection concluded after 13 interviews with 14 participants as the research team deemed sufficient informational power was attained (Malterud et al., 2016). The interviews ranged from 36 to 67 min (mean: 57 min). Participants were entered into a raffle to win one of four $100 checks as compensation.

## 2.3 | Data collection

Prior to selecting interview participants, a consent form and a demographic survey were distributed via LimeSurvey to both ensure eligibility and diversity among the respondents. The demographics survey was designed to gather information about their child's intersex variation, time elapsed since diagnosis, sex assigned at birth, gender identity, as well as the parents' ages, racial and ethnic backgrounds, and educational levels. The semi-structured interview guide, developed by our research team, underwent a pilot test involving a parent of an intersex child and another parent of a child with Down syndrome. The interviews were conducted by one of the first authors who, at the time, was a graduate student in genetic counseling and is queer. Participants were assured they could skip questions or end the interview at any time, emphasizing their autonomy. To maintain reflexivity and mitigate potential subjectivity and biases in the interview process and subsequent data interpretation, the interviewer kept a journal to record her reflections (Braun & Clarke, 2006, 2021).

The semi-structured interview guide encompassed questions exploring four domains: (1) experiences related to discovering their child's intersex variation; (2) encounters with and access to healthcare services for their child's care; (3) perspectives concerning psychosocial challenges faced by the intersex community; and (4) insights about genetics professionals' role in promoting affirming healthcare. This paper's analysis concentrates on the second and fourth domains.

## 2.4 | Data analysis

The interviews conducted through Zoom were audio-recorded, transcribed, and de-identified by a third-party transcription service. To manage and analyze the data, we employed NVivo Version 20.6.1, a qualitative research tool. Drawing from the theoretical underpinnings of queer theory (Watson, 2005) and an agency-based model of intersex health (Crocetti et al., 2020), we utilized reflexive thematic analysis, as outlined by Braun and Clarke (2006, 2021). This approach allowed us to thoroughly examine the rich and multifaceted perspectives of our participants and to delve into the nuanced intersections of intersex healthcare, identity, and the evolving paradigms of medical practice.

The coding process was a collaborative effort, involving the first authors and two other members of the research team, aimed at ensuring comprehensive engagement with the dataset. To initiate the coding process, an inductive approach was employed, where six transcripts were independently reviewed by the coding team. This initial phase involved generating concepts of meaning and highlighting representative quotes across the dataset. Subsequently, one of the first authors took on the task of analyzing the remaining seven transcripts. To refine the coding framework and reconcile any discrepancies, study investigators engaged in iterative discussions and reflective processes. These collaborative efforts led to the conceptualization of themes.

# 3 | RESULTS

Fourteen parents participated in 13 interview sessions, with both parents present during Interview 8. Only one parent from the joint interview filled out the demographic survey. Participants' children spanned a broad age range, from 2 to over 20 years. They described their children's variations as the following: complete androgen insensitivity syndrome, 17-beta hydroxysteroid dehydrogenase 3 deficiency, 45X/46XY, ovotesticular disorder/difference, partial androgen insensitivity syndrome, mixed gonadal dysgenesis, micropenis, intersex, and 45X/47XXX. The time since diagnosis ranged from 1 to over 16 years, with most diagnoses received in the neonatal stage. The parents were predominantly female and married, had a range of educational backgrounds, all resided in the United States or Canada, and most participated in intersex support groups. The racial/ethnic composition of participants was predominantly White, with one participant identifying as Southeast Asian. Tables 1 and 2 provide an overview of the demographic information for the participants' children and participants, respectively.

**TABLE 1** Child demographic information.

| Age (years) | $N = 13$ |
|---|---|
| 2–5 | 4 |
| 6–10 | 2 |
| 11–15 | 2 |
| 16–20 | 0 |
| >20 | 5 |
| Child's variation (self-describe) | |
| Complete androgen insensitivity syndrome | 4 |
| 17-beta hydroxysteroid dehydrogenase 3 deficiency | 2 |
| 45X/46XY | 1 |
| Ovotesticular disorder/difference | 1 |
| Partial Androgen Insensitivity syndrome | 1 |
| Mixed Gonadal Dysgenesis | 1 |
| Micropenis | 1 |
| Intersex | 1 |
| 45X/47XXX | 1 |
| Years since diagnosis | |
| 1 | 2 |
| 2–3 | 1 |
| 4–6 | 4 |
| 7–10 | 2 |
| 11–15 | 1 |
| >16 | 3 |
| Stage at which diagnosis was received | |
| Prenatal | 1 |
| Neonatal | 8 |
| Pediatric | 4 |

*Note*: Only select demographics are illustrated to preserve anonymity of participants.

**TABLE 2** Parent demographic information.

| | |
|---|---|
| Age (years) | *N* = 13 |
| 20–24 | 1 |
| 35–39 | 3 |
| 40–44 | 2 |
| 45–49 | 3 |
| 50–54 | 1 |
| 55–59 | 2 |
| 60–64 | 1 |
| Gender | |
| Female | 12 |
| Male | 1 |
| Relationship status | |
| Married | 12 |
| Single | 1 |
| Highest level of education | |
| Some college or secondary education | 5 |
| Bachelor's degree | 2 |
| Graduate school and/or professional degree | 5 |
| Other | 1 |
| Race/ethnicity | |
| White | 12 |
| Southeast Asian | 1 |
| Country of residence | |
| United States | 9 |
| Canada | 4 |
| Member of intersex support group | |
| Yes | 12 |
| No | 1 |

*Note*: Only select demographics are illustrated to preserve anonymity of participants. Thirteen of the parents' demographics are represented, as only one member of the joint parent interview completed the demographic survey.

Four overarching themes were generated: (1) Challenges in accessing quality healthcare; (2) Genetic diagnosis as a source of clarity for parents; (3) Incremental information disclosure: Timing and sensitivity in intersex care; and (4) The family dynamic in intersex healthcare.

## 3.1 | Theme 1: Challenges in accessing quality healthcare

Participants offered numerous accounts of needing to seek quality care outside of their local health institutions. Many described traveling "to a hospital two states away" (Interview 6) or "eight hours one way" (Interview 11) in order to access HCPs they could trust: "The professionals that we do trust – our team – they're three hours away in a larger city" (Interview 2). The driving force for this was rooted in their local providers' lack of awareness of and experience with intersex variations:

> I would ask … 'How many girls like mine have you worked with?' And I would get the answer, 'Zero.' I didn't really feel comfortable having my daughter go to providers like that, which is why I ended up changing course.
>
> (Interview 1)

Further, many participants emphasized the need to "become more of an advocate" (Interview 12) for their child in healthcare settings. These efforts include advocating for respectful and sensitive healthcare: "There have been some [specialists] who used [insensitive] language. … I walk into every appointment prepared to shut that down. … Language like that was just unacceptable and it wasn't stopping" (Interview 3). These efforts also extended to advocating for access to specialized care for their child:

> I pushed for referrals to the DSD Clinic. … I've had great experiences at DSD Clinics and I wish that there were a guide, 'Here are all the DSD Clinics,' … They see a lot of [intersex] patients'. This information would be so helpful.
>
> (Interview 1)

Even for participants who did access HCPs specializing in intersex variations, most remarked that they still lacked mental health services: "I could get the services as far as an endocrinologist, a geneticist – but the emotional services, no" (Interview 12). Further, many highlighted a desire to have "more of a personal relationship" (Interview 9) with their child's HCPs, including greater access to and partnership in their child's care:

> What about just answering [my calls] when I need to talk … and ask a question? I don't care if you bill me. The fact that I have to make an appointment, which is [weeks/months] away, to get any questions answered … was very frustrating.
>
> (Interview 12)

A key recommendation from participants followed in specialty clinics related to the potential benefits of holistic communication among the care team, "so by the time they have spoken with the parents … it feels like there's more alignment" (Interview 6). Without this multidisciplinary approach, some described information to be disjointed and misleading. One participant explored the ways in which this communication would assist in greater psychosocial support:

> Having the team meet together … it wouldn't matter who's giving that [psychosocial] information. … There are definitely [pieces of information] that are

> more specific to each [specialty] but, especially the social part, I think [it] is helpful to have that team perspective.
>
> (Interview 7)

While most parents expressed that they would appreciate greater provision of psychosocially nuanced care, many felt that this conversation would be best led by HCPs with whom they "see more often" and have "that relationship, that rapport with [them] that we felt more comfortable" (Interview 7). Multiple parents attested that this responsibility often falls to endocrinologists, calling them "the quarterback of the DSD team" (Interview 1) and describing endocrinologists as "best equipped and [having] the best experience with all of that [psychosocial care]" (Interview 6). Parents emphasized the importance of endocrinologists' training and preparation to navigate such sensitive conversations: "It will be good for the endocrinologist to learn as much as they can because they tend to be the main doctors that intersex people look for" (Interview 5).

When asked whether it would be helpful or appropriate for genetics professionals to explore psychosocial concerns, such as societal norms surrounding sex/gender, all participants expressed a preference to unpack these issues with someone who has experience and expertise:

> Part of me wants to say the more reassurance and the more concern for our child the better, but I think it wouldn't be as appropriate [for genetics to explore psychosocial topics] unless there was more … experiential training and communication. … I wouldn't want it as a parent if they weren't bathed in specialization or experience.
>
> (Interview 6)

## 3.2 | Theme 2: Genetic diagnosis as a source of clarity for parents

Participants evidenced the important role genetics has in offering clarity and comfort by establishing a clinical diagnosis. One participant explained: "I actually found out [about the diagnosis from] the genetic counselor. … So I feel like I got my answer. … Once I know the cause, I can figure out how to handle it" (Interview 1). Another participant explored how "the confirmation of a diagnosis is also helpful in … thinking about what the future could entail" (Interview 7). A sensitive approach to this diagnostic process was emphasized: "Be as transparent as possible. … Take the time, answer the questions, don't put any blame and just talk about outcomes. … If I go back to that day, the number one thing that made me feel OK was the geneticist" (Interview 10). Some participants considered how access to diagnostic information via prenatal genetic testing allowed for greater preparation to welcome their future child:

> Because I did have access to that pre-testing while still being pregnant, we have just had much more time to mentally prepare and to actually gather resources and get informed. … Having that pre-knowledge of what she had and then being able to act on that has been pretty significant.
>
> (Interview 9)

In contrast, some participants explored how a lack of access to a diagnosis led them to feel defenseless against the uncertainty of their child's future: "Early on, it was more like it could be anything" (Interview 6). A duality can be drawn with one participant who felt the identification of an underlying genetic cause was less of a priority compared to the importance of clarifying the larger health implications for their child:

> I cared more about the health of my child and what we were supposed to do now. … [It was] frustrating that I got my geneticist appointment before I got my appointment with my endocrinologist, or any other doctors. Because I was like, "This doesn't matter to me right now."… That could have waited.
>
> (Interview 12)

In an effort to establish a clinical diagnosis, some genetics centers may test for a broad range of VSC-related genes, while others may opt for a tiered testing approach. One parent advocated for equitable access to comprehensive genomic testing, considering the burden of undergoing multiple tiered tests and the considerable phenotypic overlap between VSCs:

> In the [United] States, because of insurance issues they choose a more [targeted test]. … If there was a way for the healthcare system to do the 70 [gene] panel … because so many of the variations have a lot of similarities. So that would have been helpful instead of just getting blood test after blood test.
>
> (Interview 7).

Finally, most participants expressed a desire for more contact and follow-up with their genetics centers: "I wish we had more opportunities to talk to them, to see them and to meet with them" (Interview 9).

## 3.3 | Theme 3: Incremental information disclosure: Timing and sensitivity in intersex care

Consistent across parental accounts of their child's diagnostic experience was the need for HCPs to consider the pace at which information is given to families. Many participants discussed the excess information provided and expressed how dosing and revisiting this guidance across multiple sessions can help to integrate it: "It's hard

to retain information. … You just need to go more than once to fully get all the information" (Interview 13).

Many parents conveyed the difficulty of absorbing the information within a single visit:

> I don't remember half of what they said at those appointments. … I wish sometimes they'd reconnect after an appointment and say, 'Hey have you thought about … what can we help you with again?' Because it's information overload.
>
> (Interview 12)

The desire for more repetition and follow-up with healthcare teams was echoed in relation to psychosocial topics, such as discussions regarding sexual orientation or gender identity, particularly because HCPs provide information "from a medical point of view, [which] can be overwhelming for parents" (Interview 5). Instead, this participant suggested checking in "a few sessions after the initial shock … [or] for the geneticist to meet the parents once a year … [because] they said, 'Oh yeah, no fallopian tubes so can't have kids.' And then 'cancer.' So I think that's all a shock a parent can deal with, so this non-binary, I think it might be too early. … And then the definitions might get them confused" (Interview 5).

There was variability in parental perspectives regarding which information should be provided immediately versus over time. As part of this, some parents suggested that HCPs can play a crucial role in demystifying sex and gender, underscoring the idea that HCPs should not only offer medical guidance but also support in navigating topics with empathy and sensitivity:

> It's important [to] get out of the idea that you have to be a perfect male or a perfect female to be acceptable. A genetic doctor can help framework that information [and] put a lot of parents at ease. … I find that with genetic counselors, they very much stay in their [medical] lane and not necessarily the social side. … It would be nice for people to know that there is more flexibility in the actual characteristics that can present.
>
> (Interview 9)

One participant recounted their HCPs' nuanced and thoughtful discussions about sex and gender on their newborn's first day of life, in which they explored sex assignment and potential future gender fluidity:

> The geneticist was the only one who said anything about [sex assignment vs. gender fluidity]. … She was very helpful. … She [said] we should always be prepared to offer all types of toys to our child. … And that … by age two or three our child would likely be gravitating towards certain things. … Then the surgeon chimed in that an intersex child may always kind of go-between and … to just be prepared for that.
>
> (Interview 3)

### 3.4 | Theme 4: The family dynamic in intersex healthcare

Across interviews, participants underscored the profound influence genetic investigations have on their family dynamics. This was exemplified through the need for participants to engage family members to elucidate family history information and the accompanying unease experienced when initiating and navigating these discussions with their relatives:

> We were told to ask a lot of questions from family, and look for … women [with infertility], … or maybe a man who urinated out of the side of their penis. … Like I'm going to ask somebody that. It just doesn't come up in conversations. I couldn't even [ask] because then they're going to go, 'Why are you asking this question?' … I don't know how to find the information without sharing my daughter's privacy.
>
> (Interview 12)

Conversely, for some, the recognition of parallels between their child's and their relatives' history introduced a sense of empowerment. One participant expressed the benefit of having other intersex family members for their child to turn to since the variation "ran in our family … she's always had those mentors too" (Interview 4). This familial representation facilitated the integration of diagnostic information into their family's collective identity:

> Having … two adult cousins that have the same intersex variation – which they didn't even know their diagnosis until after our son was born – … we had a little bit less questions for our geneticist because we already had that family link.
>
> (Interview 3)

Recurrent throughout discussions with participants was the experience of guilt and shame as they progressively revealed genetic indicators of intersex variations within their family lineage. This genealogical investigation was noted for its downstream impacts on other family members once the underlying genetic cause was identified: "As a mother [there] was a whole lot of acceptance and work around not feeling guilty. My mom felt guilty and responsible for passing it onto me and not knowing and … suddenly we were able to kind of track way back into our family history" (Interview 3). Several participants described a sense of intrusion as they uncovered potential diagnoses in their own relatives before those individuals were made aware:

> It was tricky because I knew the possible diagnosis even before my [relatives]. … I had to figure out a way to share what I knew. … I didn't feel like I was the right one to share the information but after my genetic counseling I said, 'I've learned some things and here's a contact to get some more information about it if you want. I found [that] really tricky because none of the doctors told them at any point about diagnoses.
>
> (Interview 4)

A common sentiment among participants was the need for healthcare providers to adopt a holistic approach that considers the needs and well-being of the entire family, rather than exclusively directing their care to the child. Some emphasized the importance of this family-centered approach as a means of addressing the family's grief: "Especially early on [parents are] … grieving for themselves in addition to grieving for their child" (Interview 4). Some participants also stressed the value of this comprehensive approach in acknowledging and coping with the guilt experienced by parents: "Yes, the child's going through it, but the parents also feel guilty because no one wants their child to hurt and, also, the reason why our child is [intersex] is something in our genetics. … You're counseling both people" (Interview 11). Many parents postulated the value of healthcare teams treating the family as a collective unit:

> Giving [families] names of other families they can talk to and referring to them to books and education on the internet that's appropriate. … Treat the patient's family, the psychosocial part, and the physical part, treat the entire thing.
>
> (Interview 8)

## 4 | DISCUSSION

The findings presented in this study shed light on the experiences and challenges faced by parents of intersex children. Through an analysis of the themes, we can gain deeper insights into the issues surrounding intersex healthcare and potential avenues for improvement.

### 4.1 | Enhancing intersex healthcare access, diagnostics, and support

Accessibility to quality intersex healthcare poses a substantial challenge for parents, as participants described often requiring extensive travel to access experienced providers or specialized care centers. This parallels the challenges faced by both the broader rare disease and transgender communities (Belzer et al., 2022; Budych et al., 2012; Zatloff et al., 2021). Epistemic injustice, which involves the unfair distribution of knowledge and understanding, is intertwined with the challenges faced by intersex individuals and their families (Fricker, 2007; Haghighat et al., 2023). The prevailing lack of awareness and understanding in both the medical community and society at large about intersex variations perpetuates epistemic injustice (Crocetti et al., 2020; Liang et al., 2017). It manifests as medical professionals failing to provide comprehensive and respectful information, partly due to insufficient training, which results in a knowledge gap on intersex issues (Crocetti et al., 2020; Jaramillo et al., 2019). Consequently, parents may not receive the necessary support and guidance, leaving them to navigate complex healthcare systems with limited information (Jaramillo et al., 2019). Epistemic injustice deprives individuals with VSCs of the confidence to question their treatment, fostering a perception of powerlessness and restricted agency over their bodies (Jones, 2022). Parents frequently find themselves in an advocate role for their child's healthcare needs, which introduces its own set of challenges, including the necessity for a high level of health literacy and the need to navigate healthcare systems that do not consistently demonstrate respect or sensitivity to their needs (Belzer et al., 2022; Crocetti et al., 2020; Jaramillo et al., 2019; Weidler & Peterson, 2019). To ameliorate this gap, healthcare organizations should prioritize the expansion of local, specialized intersex healthcare services and invest in VSC training programs for healthcare professionals.

The diagnostic process can play a crucial role in intersex care and can affect parents in their care of their children. Participants elucidated the benefits provided by genetic diagnostic clarity, including the comfort offered by receiving a concrete understanding of the cause and implications of their child's variation. This understanding enables parents to make more informed decisions about their child's healthcare and well-being, which can reduce uncertainty and distress (Jaramillo et al., 2019; Jones, 2022; Lundberg et al., 2017). Furthermore, genetic diagnoses inform patient-tailored care plans, which can lessen ambiguity and facilitate a more effective and patient-centered approach to care (McCauley, 2017; Simpson et al., 2018). In alignment with prior findings, our research attests that the earlier disclosure of diagnostic information granted by prenatal genetic testing may permit greater familial preparedness, both in terms of mental readiness and the coordination of appropriate medical care (Jaramillo et al., 2019). Our research contributes new insights into the value of ensuring equitable access to comprehensive genomic testing for this patient population. This approach would help to mitigate the psychological and economic burdens of tiered genetic testing as well as the extensive diagnostic odyssey, in parallel to rare disease literature (Lavelle et al., 2022). Importantly, diagnostic testing should never be ordered under the guise of a "social emergency" to identify an individual's "true sex," as this rhetoric perpetuates historical malpractice, pathologization, and interphobia (Crocetti et al., 2020; Prandelli & Testoni, 2020; Raz, 2023).

It is essential to recognize the potential discrepancy in perspectives and priorities between intersex individuals and their parents when it comes to genetic testing and diagnosis for intersex variations. As described in this study, parents may prioritize obtaining a diagnosis to understand their child's condition and future, while intersex children, as they mature, may focus on issues like

self-acceptance, identity, peer support, and autonomy in healthcare decision-making (Haghighat et al., 2023; Jones, 2022). This divergence underscores the need for a patient-centered approach that acknowledges the distinct perspectives and needs of parents as well as intersex children within the healthcare system, ultimately prioritizing the children's best interest and autonomy (Crocetti et al., 2020; Dickens, 2018; Jones, 2022). From an ethical standpoint, priority should be given to genetic testing that could impact necessary medical decisions and treatment, as can be the case with adrenal insufficiency sometimes seen in X-linked adrenal hypoplasia congenita (Haghighat et al., 2023; Lin et al., 2006). Genetic evaluation for the sole purpose of determining the underlying cause of intersex traits should be postponed until the individual can provide informed consent, in parallel to considerations applied to genetic testing for adult-onset conditions. This recommendation aligns with established guidelines (Committee on Bioethics, Committee on Genetics, and American College of Medical Genetics and Genomics Social, Ethical, and Legal Issues Committee, 2013; National Society of Genetic Counselors, 2018).

Moreover, concerns have been raised regarding the potential negative consequences of genetic testing for traits that may be integral to an individual's identity, such as autistic or transgender identities. These concerns include fears of stigmatization or discrimination, with the argument that these traits should be embraced as part of the broader spectrum of human diversity rather than pathologized or subjected to genetic investigation (Byres et al., 2023; Dickens, 2018; Rajkovic et al., 2022). To our knowledge, similar studies have not been conducted within the intersex community, though previous accounts from intersex individuals have highlighted the need to depathologize intersex traits (Haghighat et al., 2023; Jones, 2022). The dynamics of medicalization are intricate, as shared diagnoses can facilitate connections for some people with VSCs, while others may experience isolation due to the stigma of pathologization, rigid diagnostic criteria, or sociomedical expectations regarding treatment paths (Jones, 2022). An important balance needs to be drawn between normalizing intersex traits and affirming the diverse identities of individuals with VSCs while also addressing any potential medical needs or co-occurrences (Haghighat et al., 2023; Wang et al., 2023).

The multidisciplinary approach to intersex healthcare has proven invaluable, with holistic communication among healthcare team members being essential (Gramc et al., 2021; Lee et al., 2016; Roen, 2019). Participants in this study identified endocrinologists as central figures in intersex care, which highlights the need for specialized training that includes sensitive navigation of psychosocial concerns. Similarly, genetics professionals can play a unique role in addressing psychosocial topics. To meet this need, experiential training and resources created by community-led organizations should be integrated into health professionals' education (Prandelli & Testoni, 2020; Weidler & Peterson, 2019). Furthermore, healthcare institutions and educational programs should incorporate interdisciplinary training to foster collaborative and effective communication among healthcare team members (Gramc et al., 2021). HCPs must be equipped to demystify discussions related to sex and gender and offer sensitive education and peer support to families (Crissman et al., 2011; interACT and Lambda Legal, 2018; Llorin & Zayhowski, 2023). Importantly, HCPs can deconstruct the binary notion of sex and gender by recognizing and communicating that genetic and chromosomal factors are not the sole determinants of sex, and that both gender and sexuality are multifaceted and distinct from sex (Llorin & Zayhowski, 2023). Further, MDTs must integrate psychosocial support into intersex care (Roen, 2019; Sandberg et al., 2017), and recent research has recommended a trauma-informed approach to intersex care given the common experience of medical harm among the intersex community (Haghighat et al., 2023; Wang et al., 2023).

The dosing of information surrounding a child's intersex variation demands a nuanced approach from the healthcare team. Participants offered varying perspectives on what information should be provided immediately versus over time, which bolsters the importance of accommodating distinct learning preferences. Genetics professionals can play an influential role in delivering this information at a comprehensible and accessible pace (Saulnier et al., 2021; Simpson et al., 2018). An acknowledgment of diverse learning needs post-diagnosis is crucial: some individuals prefer immediate details and decision-making, while others may feel overwhelmed and opt for a gradual approach (McCauley, 2017). Tailoring information to patient preferences, incorporating repetition and follow-up visits, and encouraging them to express their understanding can effectively address the potential disorientation of navigating a new diagnosis (McCauley, 2017; Roen, 2019). The development of clear guidelines, protocols, and evidence-based practices for information dosing and family support, created in collaboration with the intersex community, would not only improve quality of care but also mitigate epistemic injustice by ensuring that knowledge is shared and disseminated in an equitable manner (interACT and Lambda Legal, 2018; Jaramillo et al., 2019; Prandelli & Testoni, 2020).

The influence of sociocultural norms and societal stereotypes regarding sex and gender diversity has been identified as a significant barrier for intersex individuals and their families when it comes to communicating with others and seeking support (Jones, 2022; Lundberg et al., 2021; Weidler & Peterson, 2019). The impact of healthcare providers framing intersex variations within a binary sex/gender construct due to sociocultural biases perpetuates unequal care and maladaptive coping behaviors (Roen, 2019; Wolfe-Christensen et al., 2014). This study underscores the significance of addressing the silence and misconceptions surrounding intersex variations and highlights the need for greater acceptance in society. Several calls have advocated for the inclusion of intersex topics in sexuality education, with the aim of promoting visibility and awareness (Lundberg et al., 2021). Moreover, VSCs need to be taught more thoughtfully in biology, genetics, and clinical education, incorporating nuanced discussions that reflect the complexity and diversity of intersex experiences (Lundberg et al., 2021; Prandelli & Testoni, 2020). Systematically reforming educational practices can mitigate the gender and genetic essentialism that underlie misconceptions and discrimination (Llorin & Zayhowski, 2023).

### 4.2 | Family dynamics and collective well-being

This study demonstrates that family dynamics are influenced by genetic investigations, extending beyond the immediate family unit, in alignment with previous research (Simpson et al., 2018; Srinivasan et al., 2020). These findings enhance our understanding of how engaging with relatives for family history information impacts family dynamics in the context of intersex health. Parents' narratives of shame and guilt in response to uncovering genetic indicators of intersex variations within their family lineage are noteworthy. Stigma surrounding intersex variations plays a central role (Crissman et al., 2011; Sandberg et al., 2017; Wisniewski, 2017), as parents described this as a substantial barrier that dissuades them from engaging in transparent dialogues with family members regarding family history, genetics, and their child's intersex variation. The pervasive fear of judgment, prejudice, or misconceptions surrounding these variations hinders parents from sharing information with their extended families (Lundberg et al., 2021; Roen, 2019; Weidler & Peterson, 2019), even when those families may already include individuals with similar variations. Participants described how this reluctance to engage in open conversations not only enhanced parents' feelings of isolation but also hindered the formation of support networks. In the pursuit of discovering the genetics underlying their child's intersex variation, some parents described an ability to deduce other potentially affected family members, and expressed a sense of intrusion into their family's privacy. These reflections highlight the far-reaching consequences of societal stigma and underscore the urgent need to foster more inclusive and compassionate discussions surrounding intersex topics within and beyond the family unit. Healthcare providers have a duty to address and combat this stigma, as this is integral to facilitating healthy communication and promoting the well-being of both the intersex patient and their family (Crissman et al., 2011; Llorin & Zayhowski, 2023).

A family-centered approach is essential to providing comprehensive intersex care, prioritizing the family as a collective unit. In the context of Western medicine, the predominant approach often leans toward individualistic care, emphasizing the treatment and management of an individual patient's health (Jones, 2022; Lee et al., 2016). However, the hereditary nature of intersex variations challenges this paradigm, as is true with many genetic conditions (Simpson et al., 2018; Srinivasan et al., 2020). In fact, literature has suggested that genetic counselors have an ethical obligation to consider their patients within a wider family context (Jamal et al., 2020). As a result, collectively thinking about family health becomes pivotal in the care of intersex individuals. This paradigm shift acknowledges that intersex conditions have an impact not only on the individual affected but also on their immediate and extended family members. Understanding and addressing this broader perspective is vital in providing holistic support and healthcare that meets the psychosocial and emotional needs of intersex families (Crocetti et al., 2020; Gramc et al., 2021; Weidler & Peterson, 2019). Moreover, the benefits of connecting their patients with other intersex families should be recognized by HCPs (Jaramillo et al., 2019; Jones, 2022). Recognizing the interdependence of family health is essential to offering the most effective and compassionate care to those with intersex variations.

### 4.3 | Limitations

Several limitations should be considered when interpreting the findings of this study. First, most parents interviewed for this project have previously connected with intersex support groups, which may limit the generalizability of the results to a broader population of parents of intersex children that may not have had this opportunity. The narrow demographics affect the generalizability of the findings to a more diverse population of intersex families with varying demographic and cultural backgrounds; individuals with marginalized identities are likely to face additional obstacles. The study also focused on parental perspectives, and further research should include the voices of intersex individuals themselves. While parents can serve as crucial advocates and support systems for their children, their experiences and priorities can diverge from those of the intersex individuals they care for. Moreover, recruitment advertisements employed the terminologies “intersex” and “variations of sex characteristics.” Consequently, the perspectives and insights shared by study participants may not reflect those of families who identify with alternative terminologies.

### 4.4 | Future directions

Several avenues should be explored to advance our understanding of intersex family experiences in genetic care. There is a pressing need for comprehensive investigations into the challenges faced by intersex individuals and their families across diverse cultural and socioeconomic backgrounds. Such studies can shed light on the intersectionality of experiences and help tailor interventions to specific contexts. In addition, further investigation is required to understand the potential divergence of perspectives and priorities between individuals with VSCs and their parents when making healthcare decisions. Moreover, research should focus on the long-term outcomes and psychosocial well-being of intersex individuals who underwent genetic testing and care during childhood. Longitudinal studies can assess the impact of early interventions on their quality of life, mental health, and self-identity in adulthood. Comparative research across different healthcare systems and countries can highlight best practices and disparities in genetic care for intersex individuals. These studies can identify structural and systemic factors that contribute to or hinder equitable access to quality care. Additionally, investigations into the role of advocacy and support groups in shaping family experiences within genetic care are essential. Understanding the ways in which these groups influence decision-making, emotional well-being, and information-sharing can inform strategies for better support. Research should focus on assessing the effectiveness of educational interventions for genetics professionals. Studies that

evaluate the impact of training programs on provider knowledge, attitudes, and practices can contribute to more informed and patient-centered care.

## 5 | CONCLUSIONS

This study provides important insights into the experiences of and challenges faced by parents of intersex children as they navigate the complex landscape of intersex healthcare. These findings shed light on the need for improved accessibility, diagnostics, and support in intersex healthcare, emphasizing the role of genetics professionals and healthcare institutions in promoting comprehensive and patient-centered care. The study highlights the benefits of genetic diagnosis and the necessity for a patient-centered approach that respects the autonomy and perspectives of both parents and intersex individuals. The multidisciplinary approach to intersex healthcare and the integration of psychosocial support are essential components in addressing the multifaceted needs of intersex families. Additionally, the study emphasizes the significance of information dosing and tailored family support to ensure equitable dissemination of knowledge and mitigate epistemic injustice. Addressing these challenges requires collaboration among healthcare organizations, educational programs, and advocacy groups to shape more inclusive, patient-centered healthcare. In the pursuit of healthcare that respects and uplifts the diverse experiences and identities of intersex individuals and their families, it is our collective responsibility to promote understanding, advocate for change, and ensure that no one is left behind in their journey toward equitable and compassionate healthcare.

### AUTHOR CONTRIBUTIONS

**Kayla Horowitz:** Conceptualization, Methodology, Formal analysis, Investigation, Data curation, Writing—original draft, Writing—review and editing, and Project administration. **Kimberly Zayhowski:** Conceptualization, Methodology, Formal analysis, Writing—original draft, Writing—review and editing, Supervision. **Nicole Palmour:** Conceptualization, Funding acquisition, Formal analysis, Writing—review and editing. **Darius Haghighat:** Conceptualization, Formal analysis, Writing—review and editing. **Yann Joly:** Conceptualization, Methodology, Funding acquisition, Formal analysis, Writing—review and editing, Supervision, Project administration. Kayla Horowitz and Kimberly Zayhowski have made equal contributions to this work and should be regarded as joint first authors.

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### CONFLICT OF INTEREST STATEMENT

Kayla Horowitz, Kimberly Zayhowski, Nicole Palmour, Darius Haghighat, and Yann Joly declare that they have no conflicts of interest.

### DATA AVAILABILITY STATEMENT

Research data are not shared due to remote risk of re-identifying a participant.

### ETHICS STATEMENT

Human studies and informed consent: This study was approved by the McGill University Health Centre (MUHC) Research Ethics Board [2022-7864] in November 2021. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Informed consent was obtained from all participants prior to their inclusion in this study.

Animal studies: No non-human animal studies were carried out by the authors for this article.

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