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Genetic Counselors' Experiences with and Approaches to Discordant Genotypic and Phenotypic Sex Detected via Non-Invasive Prenatal Testing

Emily Stiglich

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GENETIC COUNSELORS’ EXPERIENCES WITH AND APPROACHES TO DISCORDANT GENOTYPIC AND PHENOTYPIC SEX DETECTED VIA NON-INVASIVE PRENATAL TESTING

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GENETIC COUNSELORS’ EXPERIENCES WITH AND APPROACHES TO DISCORDANT GENOTYPIC AND PHENOTYPIC SEX DETECTED VIA NON-INVASIVE PREGNATAL TESTING

A
THESIS
Presented to the Faculty of
The University of Texas
MDAnderson Cancer Center UTHealth
Graduate School of Biomedical Sciences
in Partial Fulfillment
of the Requirements
for the Degree of
MASTER OF SCIENCE

by
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Houston, Texas
May 2020
GENETIC COUNSELORS’ EXPERIENCES WITH AND APPROACHES TO DISCORDANT GENOTYPIC AND PHENOTYPIC SEX DETECTED VIA NON-INVASIVE PRENATAL TESTING

Emily Catherine Stiglich, BS

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As the use of non-invasive prenatal testing becomes more ubiquitous during pregnancy, genetic counselors (GCs) will see clients more frequently for discordant sex identification via non-invasive prenatal testing (NIPT-DSI). Thus, it is imperative to investigate what GCs consider important when counseling about NIPT-DSI and assess how GCs perceive their role in such cases. Prenatal and pediatric GCs were surveyed regarding previous experiences of NIPT-DSI, comfort levels of topics relating to NIPT-DSI, and perceived importance of potential discussion topics in a counseling session ($n = 108$). The survey consisted of two vignettes, presenting cases of NIPT-DSI identified prenatally in one scenario and postnatally in the other scenario. Sixty-nine percent of GCs surveyed reported past experiences with NIPT-DSI ($n = 75$), with 74% of GCs that see prenatal patients ($n = 64$) and 45% of GCs that work in pediatrics ($n = 22$) reporting such experiences. GCs generally expressed comfort regarding the discussion of differential diagnosis, discordant NIPT results, sharing information with others, and recurrence risk, but discomfort regarding the discussion of genital surgeries. GCs ranked discussion of differential diagnosis, testing options, and parental support as the most important topics for an initial NIPT-DSI genetic counseling session. Additionally, while most participants indicated that they would offer genetic testing and referrals to other specialties in the hypothetical scenarios, there was little agreement between counselors of which testing and referral options to include. Overall, these results indicate GCs do encounter NIPT-DSI cases in their clinical practice, GCs may feel uncomfortable discussing certain topics that may arise in a session, such
as genital surgeries, and there are inconsistencies in the way that GCs would approach similar cases of NIPT-DSI. Therefore, targeted education for GCs regarding NIPT-DSI and related topics, as well as practice guidelines specific to GCs for cases of NIPT-DSI may help ensure that patients with similar indications are receiving the same quality of care.
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ABBREVIATIONS

GC = Genetic Counselor

NIPT = Non-Invasive Prenatal Testing

NIPT-DSI = Discordant Sex Identification via Non-Invasive Prenatal Testing

DSD = Differences/Disorder of Sex Development

NSGC = National Society of Genetic Counselors

ABGC = American Board of Genetic Counseling
INTRODUCTION

Differences/disorders of sex development (DSDs) are a group of conditions collectively defined as atypical development of chromosomal, gonadal, or anatomical sex (Hughes et al., 2006). DSD has been estimated to occur in one in 5500 births (Sax, 2002). Historically, individuals diagnosed with DSDs in early infancy have been detected by presence of ambiguous genitalia at birth or through abnormal newborn screening for conditions such as congenital adrenal hyperplasia (Lee et al., 2016). Another common scenario for diagnosis in older children has included abnormal pubertal development, such as primary amenorrhea or delayed or incomplete puberty (Hughes et al., 2006). However, information about fetal sex chromosomes can now be derived from newer forms of prenatal genetic screening, such as non-invasive prenatal testing (NIPT). NIPT fetal sex detection makes identification of discordant genotypic and phenotypic sex possible prior to or soon after delivery, when results are compared either to fetal sonography prenatally or physical exam postnatally (Richardson et al., 2017).

NIPT analyzes cell-free fetal DNA circulating in the maternal bloodstream and can detect fetal sex with a sensitivity of greater than 98% (Bianchi et al., 2012; Mackie et al., 2017; Pergament et al., 2014). The detection of Y chromosome material is routinely used for sex determination as early as 9 weeks gestation. It is estimated that discordance between presumed fetal sex based on NIPT results and ultrasound anatomy of genitalia occurs in as many as 1 in 1845 pregnancies (Richardson et al., 2017). As NIPT becomes more widespread in prenatal care, it is reasonable to suspect that the detection of discordant genotypic and phenotypic sex will increase.

Causes of discordant sex identification between NIPT results and fetal ultrasonography or postnatal physical exam, hereafter referred to as NIPT-DSI, depend on multiple factors and
include possibilities ranging from laboratory error to genetic conditions. The identification of discordant sex and suspected DSDs, while not typically a life-threatening occurrence, is often considered a social emergency and warrants consultation by various medical specialties, including genetics providers (Hughes et al., 2006). Parents’ first impressions from these encounters often persist and, as such, initial contact with the parents of a fetus or child in whom discordant sex has been identified is important. In previous studies, parents of children with DSDs report stress surrounding early experiences, gender assignment, genital surgeries, how to share information regarding their child’s diagnosis with others, and future concerns for their children (Alpern et al., 2016; Crissman et al., 2011). More than 40% of parents of children with a confirmed diagnosis of a DSD indicated a need for psychological support, and only half of those individuals received support that they perceived as adequate (Bennecke et al., 2015).

Prenatally, the evaluation of discordant sex is limited because assessment of internal genitalia is inadequate, thus a broad differential is generated, which may increase parental anxiety. In a study focusing on prenatal detection of DSDs, parents were found to feel overwhelmed, have difficulty bonding with the baby after having formed an identity of the other sex, and one mother experienced severe mental stress and suicidal ideation, further highlighting the need for psychosocial support (Finney et al., 2019).

Genetic counselors (GCs) have specialized training to provide information about genetic conditions like DSDs to patients, coordinate genetic testing to assist with diagnosis, and provide psychosocial support to patients and their family members (Resta et al., 2006). With respect to NIPT-DSI, GCs have the unique opportunity to be involved in cases where discordant sex is identified prenatally and postnatally. GCs are often among the first health care providers to interact with families after a suspected diagnosis of DSD and have expertise in both genetics
and psychosocial counseling. Therefore, GCs are ideal healthcare providers to provide clinical
information to families and assist them in adapting to new, and potentially confusing or
overwhelming information in cases of NIPT-DSI.

At this time there is little prior research on NIPT-DSI and none regarding the
experiences or counseling approaches of GCs with regard to this topic. Characterizing the
current and theoretical practices of GCs is necessary to understand what GCs consider
important when counseling clients with an NIPT-DSI indication. The current study aims to
identify if there is a consensus of best care practices for these patients across the genetic
counseling field and to assess how GCs perceive their role when counseling about NIPT-DSI.
Additionally, it is paramount to investigate if the perspectives of GCs concerning this topic align
with previously expressed needs of patients and families documented in the literature. As NIPT
use expands throughout general obstetric practice and NIPT-DSI, in turn, becomes a more
frequent occurrence, information about how GCs have served and should care for their clients
with NIPT-DSI will likely become crucial to prenatal and pediatric genetic counseling practices.
2 | METHODS

2.1 | Participants

After receiving IRB exempt status from the institutional review board at University of Texas Health Science Center at Houston Committee for the Protection of Human Subjects (HSC-GSBS-19-0529), participants were recruited between August 21, 2019 and January 1, 2020 via e-mail listservs distributed by the National Society of Genetic Counselors (NSGC) and American Board of Genetic Counseling (ABGC). A survey was distributed twice through the NSGC listserv and once through the ABGC listserv. Recruitment was open to board certified or board-eligible genetic counselors who have seen patients in a prenatal and/or pediatric setting within the last five years. Participants were offered the option to enter a raffle to win one of two survey incentives, valued at $25 each, by submitting personal e-mail addresses not linked to individual survey responses.

Participants were categorized into three main groups according to the type of patients they have seen in the past five years: GCs who exclusively see prenatal patients, GCs who exclusively see pediatric patients, and GCs who see both prenatal and pediatric patients.

2.2 | Instrumentation

An investigator-designed non-validated tool was developed with Qualtrics™ (Qualtrics, Provo, UT) and consisted of demographic questions and two vignettes with hypothetical NIPT-DSI scenarios presented in randomized order to each participant (Table 1). In each scenario, participants were asked to specify if and how often they had experience with similar cases and to select other specialties that had been involved in similar past cases at their practice. Participants also ranked nine counseling topics in order of perceived importance to that particular case. Topics included discussing if discordant sex could impact a person’s sexual
orientation or gender identity, providing diagnostic testing options and facilitating decisions about genetic testing, discussing possible diagnoses and their associated symptoms, discussion of potential medical management, discussing if discordant sex could impact gender assignment or gender rearing, identifying sources of social support or how the family can share information with others, providing parental emotional support, discussing the potential for recurrence, and discussing the potential social stigmas encountered by individuals with discordant sex or their families. They were also asked to identify factors that influenced their ranking of the counseling topics. Additionally, they identified the length of time they would spend in an initial genetic counseling session and specified how they would divide 60 minutes of this session among four thematic counseling categories (Diagnosis Information, Medical Management, Parental Support, and Social Concerns). Other questions requested participants to identify genetic testing and specialty referrals they would offer clients in both scenarios. Last, participants indicated their level of comfort regarding ten specific counseling topics (differential diagnosis, discordant NIPT results, sharing information with others, recurrence risk, terminology, medical management, gender identity, gender of rearing, sexual orientation, and genital surgery), as well as their overall comfort with the scenarios. GCs rated their comfort using a four-point Likert scale ranging from extremely uncomfortable (1) to extremely comfortable (4).

Most items were optional, with the exception of eligibility criteria and the division of 60 minutes between four thematic counseling categories. When an individual selected “other” in response to any item, they were required to enter text to specify the selection.

2.3 | Data Analysis
Submitted surveys were deemed eligible for analysis if at least one vignette scenario was complete. Statistical analysis was completed using Stata software (StataCorp. 2013. Stata Statistical Software: Release 13. College Station, TX: StataCorp LP). Descriptive statistics were used to analyze demographic information and responses to individual survey questions. Wilcoxon signed-rank, McNemar, and paired t-tests were used to analyze potential differences between prenatal and pediatric scenario responses. Distribution of non-paired data was assessed across categories using Wilcoxon rank-sum, One-way ANOVA, Fisher’s exact, and Kruskal-Wallis with post-hoc Dunn’s test. Statistical significance was assumed at a Type I error rate of 5% for all statistical tests.

<table>
<thead>
<tr>
<th>Scenario</th>
<th>Vignette</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prenatal</td>
<td>You are a prenatal counselor counseling a pregnant woman for discordant NIPT and ultrasound findings. This is her first pregnancy. The patient’s NIPT results are consistent with male fetal sex, but today’s anatomy scan revealed genitalia consistent with a female fetus. No other abnormalities were identified.</td>
</tr>
<tr>
<td>Pediatric</td>
<td>You are a pediatric genetic counselor asked to perform a consultation for a newborn. This is the family’s first child. NIPT results during the pregnancy indicated male fetal sex, but on physical exam, the infant’s genitalia appear consistent with female sex. No other abnormalities are present and the baby’s current health is otherwise unremarkable.</td>
</tr>
</tbody>
</table>

**Table 1.** Prenatal and Pediatric Scenarios. Both vignettes consisted of a scenario involving a fetus with NIPT results suggestive of male sex and ultrasound or postnatal physical exam consistent with a female sex.
3 | RESULTS

3.1 | Sample Demographics

Figure 1. Breakdown of Participant Responses.
One hundred forty-nine respondents took the survey. Of these, 41 had only filled out the demographics. Ninety-one participants completed all parts of the survey, whereas 17 completed the demographics and only one of the two scenarios.

A total of 149 surveys were submitted, with 108 being deemed eligible for data analysis (Figure 1). An accurate response rate cannot be determined because it is unclear how many unique GCs were reached for study recruitment by electronic advertisement.

Table 2. Sample Demographics (n = 108)

<table>
<thead>
<tr>
<th>Variable</th>
<th>n</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Type of Patients Seen in the Last Five Years</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Both Prenatal &amp; Pediatric</td>
<td>28</td>
<td>26</td>
</tr>
<tr>
<td>Pediatric only</td>
<td>21</td>
<td>19</td>
</tr>
<tr>
<td>Prenatal only</td>
<td>59</td>
<td>55</td>
</tr>
<tr>
<td><strong>Gender</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Female</td>
<td>99</td>
<td>92</td>
</tr>
<tr>
<td>Male</td>
<td>8</td>
<td>7</td>
</tr>
<tr>
<td>Prefer not to respond</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td><strong>Race &amp; Ethnicity</strong></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

1
Asian  6  6
Asian Indian  1  1
Black or African American  2  2
Hispanic  1  1
Prefer Not to Respond  1  1
White or Caucasian  101  94

Work Setting\(^1\)

<table>
<thead>
<tr>
<th>Work Setting</th>
<th>n</th>
<th>%</th>
<th>n</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Diagnostic Laboratory</td>
<td>8</td>
<td>7</td>
<td>7</td>
<td>7</td>
</tr>
<tr>
<td>Other</td>
<td>5</td>
<td>5</td>
<td>5</td>
<td>5</td>
</tr>
<tr>
<td>Physician Private Practice</td>
<td>7</td>
<td>7</td>
<td>7</td>
<td>7</td>
</tr>
<tr>
<td>Private Hospital/Medical Facility</td>
<td>27</td>
<td>25</td>
<td>25</td>
<td>25</td>
</tr>
<tr>
<td>Public Hospital/Medical Facility</td>
<td>24</td>
<td>22</td>
<td>22</td>
<td>22</td>
</tr>
<tr>
<td>University or Academic Medical Center</td>
<td>58</td>
<td>54</td>
<td>54</td>
<td>54</td>
</tr>
</tbody>
</table>

\(^1\) Four participants (3.7\%) selected multiple responses when designating race and ethnicity. Nineteen participants (18\%) selected multiple responses when designating work setting.

In the last five years, more than half of the participants have seen prenatal patients exclusively (n = 59, 55\%), approximately one fifth have seen pediatric patients exclusively (n = 21, 19\%), and about one-fourth have seen both prenatal and pediatric patients (n = 28, 26\%).

Additional demographic information collected from participants is shown in Table 2.

### 3.2 GC Experiences with NIPT-DSI

Table 3. Have GCs Counseled Cases Similar to NIPT-DSI in the Past?

<table>
<thead>
<tr>
<th>Previous Experience</th>
<th>n</th>
<th>%</th>
<th>n</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Prenatal Scenario</td>
<td>Pediatric Scenario</td>
<td>Prenatal Scenario</td>
<td>Pediatric Scenario</td>
</tr>
<tr>
<td>Yes, Once</td>
<td>22</td>
<td>25</td>
<td>8</td>
<td>16</td>
</tr>
<tr>
<td>Yes, More Than Once</td>
<td>42</td>
<td>48</td>
<td>14</td>
<td>29</td>
</tr>
<tr>
<td>No</td>
<td>23</td>
<td>27</td>
<td>27</td>
<td>55</td>
</tr>
<tr>
<td>Total</td>
<td>87</td>
<td>100</td>
<td>49</td>
<td>100</td>
</tr>
</tbody>
</table>
Although nearly three-fourths of GCs who see any prenatal patients reported having experience with cases similar to the NIPT-DSI prenatal scenario (n = 64, 74%), less than half of all GCs who see any pediatric patients (n = 22, 45%) have experience with cases similar to the NIPT-DSI pediatric scenario (p = 0.004) (Table 2a). Of those who reported previous experience with multiple cases, the majority encountered NIPT-DSI more often than once per year, but less than once per month in both the prenatal and pediatric scenario (n = 29, 74%; n = 10, 72%) (Figure 2). In the prenatal scenario, GCs who work in public hospitals reported less frequent experiences than those who do not work in public hospitals (p = 0.049), while those working in a university or academic medical setting reported more frequent experiences than those who do not (p = 0.031). There was no significant relationship between frequency of NIPT-DSI experience and work setting in the pediatric scenario. Additionally, there were no significant differences when stratified by participants’ self-reported genetic counseling specialties.

![Frequency of Similar Experiences with NIPT-DSI](image-url)
3.3 | GC Perception of Topic Importance for NIPT-DSI Sessions

Figure 3. GC Ranked Importance of Counseling Topics in Presented Vignettes. Participants ranked the topics from most important (1) to least important (9). The median rank score for topics provided by all participants is shown. Statistical analysis using Wilcoxon signed-rank test compared responses between the scenarios. **p < 0.01

**Ranking of Counseling Topic Importance**

Participants ranked discussing the differential and testing options as the two most important counseling topics in both the prenatal and pediatric scenarios when ranking the importance of nine different topics on a scale from most important (1) to least important (9). Conversely, discussing potential social stigmas and recurrence risk were ranked as least important in both scenarios (Figure 3). Half of all respondents ranked recurrence risk the same in the prenatal and pediatric scenarios; however, if discordant, participants were more likely to rank recurrence risk as less important to discuss in the prenatal scenario (69%) compared to the pediatric scenario (31%) (p = 0.006). In the pediatric scenario, GCs who exclusively see
pediatric patients ranked the discussion of sexual orientation and gender identity as less important (median = 8, IQR = 6-8) than other GCs who see prenatal patients only (median = 6, IQR = 5-7) or both prenatal and pediatric patients (median = 6, IQR = 5.5-7) (p = 0.035). GCs who exclusively see pediatric patients also ranked discussion of recurrence risk as more important (median = 8, IQR = 7-9) than GCs who see prenatal patients only (median = 9, IQR = 8-9) or both prenatal and pediatric patients (median = 9, IQR = 8-9) in the pediatric scenario (p = 0.026). Participant specialty did not influence the ranking decisions of participants in the prenatal scenario.

Factors Influencing Topic Ranking Decisions

Participants also selected factors that influenced their ranking decisions of the nine discussion topics (Figure 4). Most GCs indicated that previous experiences influenced their ranking in both the prenatal and pediatric scenarios (n = 80, 82%; n = 73, 72%, respectively). Multiple influencing factors were selected by 40 participants in the prenatal scenario (41%) and 35 participants in the pediatric scenario (34%). In the prenatal and pediatric scenarios respectively, 16% (n = 16) and 21% (n = 21) of participants selected “other” factors influenced their ranking decisions. Of participants who selected “other”, 56% (n = 9) in the prenatal scenario and 57% (n = 12) in the pediatric scenario did not select any additional factors as contributing toward their ranking decision.

Previous experiences influenced ranking more frequently for GCs who have multiple personal experiences with prenatal NIPT-DSI (n = 40, 95%) than GCs who have counseled only one similar case (n = 18, 69%), or no similar cases (n = 22, 78%) (p = 0.006). Similarly, respondents were more likely to list previous experiences as an influencing factor if they have
experience with one or more than one similar case (83%, 96%, respectively) than GCs who did not have experience with pediatric cases of NIPT-DSI (62%) \((p = 0.003)\).

While 84% of participants \((n = 76)\) selected the same influencing factors for both the prenatal and pediatric scenario, when there were differences, previous experiences were more likely to be influencing factors in the prenatal scenario \((n = 12, 80\%)\) than the pediatric scenario \((n = 3, 20\%)\) \((p = 0.035)\).

![Factors Influencing Topic Ranking Decisions](image)

**Figure 4.** Factors Influencing Topic Ranking Decisions. Percentage of participants that indicated that published guidelines, institutional policies, previous experiences, or other factors influenced their ranking decisions in the prenatal (left bars) and pediatric (right bars) scenarios. Statistical analysis using the McNemar test compared responses between the scenarios. *\(p < 0.05\)

**Duration of Initial GC Session**

Most participants indicated that they would spend between 30 and 60 minutes on an initial genetic counseling session for both the prenatal and pediatric scenario \((n = 64, 66\%; n = 59, 60\%, respectively)\) (Figure 5). Additionally, 84% of GCs reported that they would spend the same amount of time counseling in prenatal and pediatric scenario. However, if participants responded differently between the scenarios, GCs were more likely to elect spending a greater amount of time in the pediatric scenario (79%) than the prenatal scenario (21%) \((p = 0.033)\).
Participant specialty influenced the timing selection for the prenatal scenario, with GCs who exclusively see prenatal patients indicating shorter sessions than other GCs ($p = 0.018$).

**Figure 5.** Time GCs Would Spend in an Initial Session.
Percentage of participants that would spend less than 30 minutes, between 30 and 60 minutes, between 60 and 90 minutes, and greater than 90 minutes counseling in an initial prenatal (top) and pediatric (bottom) NIPT-DSI scenario.

**Duration of Time Among Four Categories**

When asked to divide 60 minutes of a hypothetical initial genetic counseling session among four thematic counseling categories, participants indicated that they would spend the most amount of time discussing diagnostic information in both the prenatal and pediatric scenario (median = 21 minutes, median = 20 minutes, respectively), and the least amount of time discussing social concerns (mean = 8.6 minutes, mean = 10.6 minutes, respectively) (Figure 6). Specialty did not influence the amount of time allotted for any category in either the prenatal or pediatric scenario. Of note, participants indicated that they would spend more time discussing social concerns and medical management in the pediatric scenario than in the prenatal scenario (mean = 10.6 minutes, IQR = 8-15 vs. mean = 8.6 minutes, IQR = 5-10, $p < 0.001$; mean = 13.1 minutes, IQR = 10-15 vs. mean = 11.1 minutes, IQR = 9-15, $p = 0.001$).
### Figure 6. Time GCs Would Spend in Each Counseling Category.
Mean and median amount of time in minutes that participants would spend in four thematic counseling categories for NIPT-DSI prenatal (upper bars) and pediatric scenarios (lower bars) out of a total of 60 minutes. Statistical analysis using a paired t-test compared responses between the scenarios. **p < 0.01; ***p < 0.001

### 3.4 GC Attitudes Toward Genetic Testing and Specialty Referrals

**Potential Genetic Testing Options in Cases of NIPT-DSI**

In both the prenatal and pediatric scenarios, the majority of participants indicated that they would offer some combination of karyotype \( n = 86, 89\% \); \( n = 86, 84\% \), respectively), FISH for the SRY gene \( n = 75, 77\% \); \( n = 75, 74\% \), respectively), and chromosomal microarray \( n = 79, 81\% \); \( n = 67, 66\% \), respectively) (Figure 7). Approximately nine percent of GCs selected “other” genetic testing in the prenatal setting, including carrier screening \( n = 2, 2.1\% \), maternal FISH for sex chromosomes \( n = 1, 1.0\% \), and a repeat of NIPT \( n = 1, 1.0\% \). The specialty of the participant did not influence their choice of testing options in either scenario. GCs who have experience with more than one prenatal NIPT-DSI case were less likely to indicate that they
would offer single gene panels than others in the prenatal scenario \((n = 13, 57\% \text{ vs. } n = 22, 30\%, p = 0.015)\). Previous experience with NIPT-DSI did not influence tests GCs would offer in the pediatric scenario. While 80\% of participants were as likely to offer chromosome microarray in both scenarios, the participants with discordant responses were more likely to offer chromosome microarray in the prenatal scenario (94\%) compared to the pediatric scenario (6\%) \((p < 0.001)\).

**Figure 7.** Genetic Testing Options GCs Would Offer Patients. Percentage of participants who selected that they would offer each of the presenting testing options for NIPT-DSI prenatal (left bars) and pediatric (right bars) scenarios. Statistical analysis using the McNemar test compared responses between the scenarios. ***\(p < 0.001\)

**Potential Referrals in Cases of NIPT-DSI**

Ninety-eight percent \((n = 95)\) and 97\% \((n = 99)\) of participants indicated that they would make at least one specialist referral in the prenatal and pediatric scenarios, respectively (Figure 8). Participant specialty influenced referral decisions in both the prenatal and pediatric vignettes. In the prenatal scenario, GCs who exclusively see prenatal patients were less likely to refer to urology (38\%) than GCs who see pediatric patients (68\%) or GCs who see both
prenatal and pediatric patients (60%) \((p = 0.035)\). In the pediatric vignette, GCs who exclusively see prenatal patients were less likely to refer to endocrinology (79%) than GCs who exclusively see pediatric patients (100%) or GCs who see both types of patients (93%) \((p = 0.035)\), while GCs who exclusively see pediatric patients were more likely to refer to urology (81%) than GCs who exclusively see prenatal patients (51%) or GCs who see both types of patients (68%) \((p = 0.046)\). Participants’ experiences with similar NIPT-DSI cases did not influence hypothetical referral recommendations.

Individual participants were overall concordant between their responses for both vignettes regarding referrals they would make as part of care for patients with NIPT-DSI. Although 75% of participants made the same referral decisions for urology in both the prenatal and pediatric scenario, when there were differences, GCs were more likely to refer to urology in the pediatric scenario than the prenatal scenario \((78\% \text{ vs. } 22\%, p = 0.011)\). Additionally, while 78% of participants made the same referral decisions regarding “other” specialties across both scenarios, when there were differences, participants were more likely to refer to “other” specialties in the prenatal scenario than the pediatric scenario \((80\% \text{ vs. } 20\%, p = 0.012)\). For the prenatal scenario, 76% of GCs \((n = 74)\) said they would refer to “other” specialties such as medical genetics \((n = 34, 35\%)\), DSD/gender clinics \((n = 11, 11\%)\), Neonatal Intensive Care Unit (NICU) or neonatology \((n = 10, 10\%)\), Maternal Fetal Medicine (MFM) or fetal center \((n = 4, 4.1\%)\), and Child Life \((n = 1, 1.0\%)\). On the pediatric side, 64% of GCs \((n = 65)\) chose to make “other” referrals, including medical genetics \((n = 31, 30\%)\), DSD or gender clinics \((n = 11, 11\%)\), Child Life \((n = 4, 3.9\%)\), and NICU or neonatology \((n = 2, 2.0\%)\).
Figure 8. Referrals GCs Would Make.
Percentage of participants that selected a specialty referral for the hypothetical NIPT-DSI prenatal (left bars) and NIPT-DSI pediatric (right bars) scenarios. Statistical analysis using the McNemar test compared responses between the scenarios. *p < 0.05

3.5 | Reported Specialties Involved in Past Cases

Figure 9. Reported Specialties Involved in Past Cases.
Percentage of participants that indicated other specialties involved in past cases of NIPT-DSI prenatal (left bars) and NIPT-DSI pediatric (right bars).
Seventy-seven percent of GCs in the prenatal vignette and 91% of GCs in the pediatric vignette, all of whom reported previous experiences with NIPT-DSI, indicated that at least one other specialty was involved in the care of their patient(s) in past cases (Figure 9). For the prenatal cases of NIPT-DSI, 68% of GCs \( (n = 44) \) said that “other” specialties, such as medical genetics \( (n = 15, 23\%) \), NICU or neonatology \( (n = 10, 15\%) \), DSD or gender clinics \( (n = 5, 7.7\%) \), and MFM or fetal center \( (n = 5, 7.7\%) \) were involved. A variety of specialties were also involved in pediatric cases of NIPT-DSI, in which 62% of GCs \( (n = 21) \) said that “other” specialties were involved, including medical genetics \( (n = 11, 32\%) \), NICU or neonatology \( (n = 4, 12\%) \), Child Life \( (n = 2, 5.9\%) \), DSD or gender clinics \( (n = 1, 2.9\%) \), MFM or fetal center \( (n = 1, 2.9\%) \), pediatric gynecology \( (n = 1, 2.9\%) \), and developmental pediatrics \( (n = 1, 2.9\%) \).

3.6 | GC Comfort with Counseling Indications of NIPT-DSI

Level of Comfort Counseling Prenatal NIPT-DSI Scenario

Overall, 87% of participants indicated that they would feel comfortable counseling the prenatal scenario (Figure 10a). The majority of participants also felt comfortable with the prospect of discussing topics such as differential diagnosis (97%), discordant NIPT (96%), sharing information with others (91%), recurrence risk (89%), terminology (67%), and medical management (65%) in the prenatal scenario (Figure 10a).

GCs who exclusively see pediatric patients were less comfortable overall with the prenatal scenario \( (\text{median} = 3, \text{IQR} = 2-3) \) than GCs who exclusively see prenatal patients \( (\text{median} = 3, \text{IQR} = 3-3) \) or GCs who see both prenatal and pediatric patients \( (\text{median} = 3, \text{IQR} = 3-4) \) \( (p = 0.001) \). Compared to participants who see only prenatal or only pediatric patients, GCs who see both prenatal and pediatric patients were more comfortable discussing gender identity \( (\text{median} = 2, \text{IQR} = 2-3 \text{ vs. } \text{median} = 3, \text{IQR} = 2-3 \text{ vs. } \text{median} = 3, \text{IQR} = 2-3, \)
respectively, \( p = 0.020 \)), gender of rearing (median = 2, IQR = 2-3 vs. median = 2, IQR = 2-3, vs. median = 3, IQR = 2-3, respectively, \( p = 0.032 \)), and genital surgeries (median = 2, IQR = 2-2 vs. median = 2, IQR = 1-3 vs. median = 2, IQR = 2-3, respectively, \( p = 0.011 \)). Additionally, respondents who exclusively see prenatal patients were less comfortable discussing medical management (median = 3, IQR = 2-3) compared to GCs who exclusively see pediatric patients (median = 3, IQR = 3-3) or GCs who see both prenatal and pediatric patients (median = 3, IQR = 3-3) (\( p = 0.017 \)). GCs who see only pediatric patients were less comfortable discussing discordant NIPT results (median = 3, IQR = 3-4) than GCs who see only prenatal patients (median = 4, IQR = 3-4) or GCs who see both prenatal and pediatric patients (median = 4, IQR = 3-4) (\( p = 0.020 \)).

GCs who have experience with more than one prenatal NIPT-DSI case expressed more comfort overall with counseling the scenario (median = 3, IQR = 3-4) compared to those who have experience with only one similar case (median = 3, IQR = 3-3), or those without any experience (median = 3, IQR = 2-3) (\( p < 0.001 \)). Compared to participants with one or more than one past experience with prenatal NIPT-DSI, GCs without experience were less comfortable discussing discordant NIPT results (median = 4, IQR = 3-4 vs. median = 4, IQR = 4-4 vs. median = 3, IQR = 3-4; \( p < 0.001 \)), differential diagnosis (median = 4, IQR = 3-4 vs. median = 4, IQR = 3-4 vs. median = 3, IQR = 3-4; \( p = 0.003 \)), and recurrence risk (median = 3, IQR = 3-4 vs. median = 4, IQR = 3-4 vs. median = 3, IQR = 3-3.5; \( p = 0.029 \)). Additionally, participants without NIPT-DSI experience were less comfortable discussing terminology than GCs who have experience with more than one prenatal NIPT-DSI case (median = 3, IQR = 2-3 vs. median = 3, IQR = 3-4) (\( p = 0.035 \)).

*Level of Comfort Counseling Pediatric NIPT-DSI Scenario*
The majority of participants (82%) indicated that they would overall feel comfortable counseling the pediatric scenario (Figure 10b). The majority of participants were also comfortable with the idea of discussing topics such as differential diagnosis (97%), discordant NIPT (93%), sharing information with others (90%), recurrence risk (91%), terminology (74%), and medical management (66%) in the pediatric scenario (Figure 10b).

Compared to GCs who exclusively see pediatric patients or GCs who see both prenatal and pediatric patients, GCs who exclusively see prenatal patients were less comfortable discussing genital surgery (median = 2, IQR = 2-3, vs. median = 2, IQR = 2-3, vs. median = 2, IQR = 1-2; p = 0.008) and medical management (median = 3, IQR = 3-3, vs. median = 3, IQR = 2.5-3, vs. median = 3, IQR = 2-3; p = 0.022).

GCs who have experience with more than one pediatric NIPT-DSI case were overall more comfortable discussing the pediatric scenario than GCs who have experience with only one case, or without any experience (median = 3.5, IQR = 3-4 vs. median = 3, IQR = 2.5-4 vs. median = 3, IQR = 3-3; p < 0.001). Additionally, GCs without pediatric NIPT-DSI experience were less comfortable discussing sexual orientation and gender identity than participants who have experience with more than one case (median = 2, IQR = 2-3 vs. median = 3, IQR = 3-4; p = 0.017; median = 3, IQR = 2-3, vs. median = 3, IQR = 3-4; p = 0.012). GCs without past experience with pediatric NIPT-DSI were less comfortable discussing differential diagnoses than participants who have experience with one or more than one similar cases (median = 3, IQR = 3-4 vs. median = 4, IQR = 4-4 vs. median = 4, IQR = 3-4; p = 0.011) and less comfortable discussing terminology than GCs who have experience with more than one similar case (median = 3, IQR = 2-3 vs. median = 3, IQR = 3-4; p = 0.048).

*Comparison of GC Comfort Between the Two Scenarios*
There was no difference in the overall comfort levels of GCs when comparing participant responses between the two scenarios.

**a. GC Comfort Levels Relating to NIPT-DSI: Prenatal Scenario**

<table>
<thead>
<tr>
<th>Topic</th>
<th>Extremely Uncomfortable</th>
<th>Somewhat Uncomfortable</th>
<th>Somewhat Comfortable</th>
<th>Extremely Comfortable</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genital Surgery</td>
<td>18%</td>
<td>55%</td>
<td>22%</td>
<td></td>
</tr>
<tr>
<td>Sexual Orientation</td>
<td>13%</td>
<td>41%</td>
<td>35%</td>
<td>11%</td>
</tr>
<tr>
<td>Gender of Rearing</td>
<td>8.3%</td>
<td>42%</td>
<td>39%</td>
<td>11%</td>
</tr>
<tr>
<td>Gender Identity</td>
<td>7.3%</td>
<td>43%</td>
<td>39%</td>
<td>11%</td>
</tr>
<tr>
<td>Medical Management</td>
<td>30%</td>
<td>50%</td>
<td>15%</td>
<td></td>
</tr>
<tr>
<td>Terminology</td>
<td>30%</td>
<td>50%</td>
<td>17%</td>
<td></td>
</tr>
<tr>
<td>Recurrence</td>
<td>7.2%</td>
<td>47%</td>
<td>41%</td>
<td></td>
</tr>
<tr>
<td>Sharing w/Others</td>
<td>7.3%</td>
<td>56%</td>
<td>35%</td>
<td></td>
</tr>
<tr>
<td>Discordant NIPT</td>
<td>31%</td>
<td>65%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Differential Dx</td>
<td>40%</td>
<td>57%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Overall</td>
<td>10%</td>
<td>64%</td>
<td>23%</td>
<td></td>
</tr>
</tbody>
</table>

**b. GC Comfort Levels Relating to NIPT-DSI: Pediatric Scenario**

<table>
<thead>
<tr>
<th>Topic</th>
<th>Extremely Uncomfortable</th>
<th>Somewhat Uncomfortable</th>
<th>Somewhat Comfortable</th>
<th>Extremely Comfortable</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genital Surgery</td>
<td>20%</td>
<td>51%</td>
<td>24%</td>
<td></td>
</tr>
<tr>
<td>Sexual Orientation</td>
<td>11%</td>
<td>34%</td>
<td>41%</td>
<td>13%</td>
</tr>
<tr>
<td>Gender of Rearing</td>
<td>41%</td>
<td>40%</td>
<td>13%</td>
<td></td>
</tr>
<tr>
<td>Gender Identity</td>
<td>33%</td>
<td>47%</td>
<td>13%</td>
<td></td>
</tr>
<tr>
<td>Medical Management</td>
<td>28%</td>
<td>49%</td>
<td>16%</td>
<td></td>
</tr>
<tr>
<td>Terminology</td>
<td>24%</td>
<td>52%</td>
<td>22%</td>
<td></td>
</tr>
<tr>
<td>Recurrence</td>
<td>7.4%</td>
<td>45%</td>
<td>46%</td>
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<tr>
<td>Sharing w/Others</td>
<td>10%</td>
<td>55%</td>
<td>35%</td>
<td></td>
</tr>
<tr>
<td>Discordant NIPT</td>
<td>7.4%</td>
<td>29%</td>
<td>64%</td>
<td></td>
</tr>
<tr>
<td>Differential Dx</td>
<td>40%</td>
<td>57%</td>
<td>17%</td>
<td></td>
</tr>
<tr>
<td>Overall</td>
<td>17%</td>
<td>65%</td>
<td></td>
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</tbody>
</table>

**Figure 10. GC Comfort Levels Relating to NIPT-DSI**

Percentage of participants who indicated that they feel extremely uncomfortable, somewhat uncomfortable, somewhat comfortable, or extremely comfortable for the scenario overall each of ten topics in the prenatal (a) and pediatric (b) scenarios.
4 | DISCUSSION

DSDs warrant multidisciplinary care from specialized healthcare providers that can provide appropriate support to parents, who may experience distress at the time of diagnosis (Hughes et al., 2006; Alpern et al., 2016). As NIPT becomes used more frequently for prenatal detection of genetic abnormalities, it is important to examine the potential secondary findings and how providers care for patients whose testing may reveal NIPT-DSI. This study utilized an investigator-designed tool to examine GCs’ previous experiences with, and attitudes toward, cases of NIPT-DSI.

4.1 | Experiences of GCs with Cases of NIPT-DSI

Most GCs have encountered NIPT-DSI in their practice. For the majority of individuals who indicated experience with more than one similar case, their experiences occurred at a frequency that is less than once per month but more than once per year. This suggests that, while NIPT-DSI may be estimated to occur once in every 1845 pregnancies, it is reasonable for GCs to encounter patients with this indication in prenatal or pediatric settings. Therefore, it is important to assess for the congruence of approaches to NIPT-DSI between GCs across the field. While GCs are not typically the only specialists involved in the care of patients with NIPT-DSI, they are often one of the first providers brought in to discuss why there may be discrepant sex identification. GCs are ideal healthcare professionals to address the multi-faceted concerns that parents of children with potential DSDs often face given the breadth of their training and current role in offering and providing prenatal or postnatal follow-up for NIPT results.

4.2 | Attitudes and Approaches of GCs toward Cases of NIPT-DSI

Compared to other topics, GCs are the most comfortable discussing differential diagnosis, NIPT-DSI, sharing information with others, and recurrence risk. On the other hand,
the majority of GCs indicated that they would feel uncomfortable discussing genital surgeries and sexual orientation with patients. Participants may feel uncomfortable discussing such topics due to level of uncertainty surrounding the information or a perceived sense that the topic is out of the scope of practice for a GC. However, GCs frequently encounter situations of uncertainty in their practice and are trained in how to discuss the limitations of knowledge with patients, thus GCs are well-equipped to discuss topics with an inherent level of uncertainty, such as sexual orientation in patients with NIPT-DSI, despite discomfort that may stem from its discussion. It is also possible that GCs expressed discomfort with these topics based on the vague verbiage used in the survey. Perhaps with a targeted question examining comfort levels discussing genital surgeries, among other topics, at a basic level, GCs would express less discomfort.

Discussing differential diagnosis, testing options, and providing support to parents were considered most important in an initial counseling session for both prenatal and pediatric scenarios. Interestingly, GCs who exclusively see pediatric patients ranked the discussion of sexual orientation and gender identity as less important than other participants. This could be explained by pediatric counselors finding other topics more important to discuss, evidenced by GCs who exclusively see pediatric patients ranking discussion of recurrence risk as more important when compared to other counselors. Differences in ranked importance of counseling topics might reflect GCs’ perceptions of what is most important to a patient at the time of the initial counseling session. For example, discussing recurrence risk may be more relevant for parents in a pediatric setting, whereas a prenatal patient may be coping with the shock and concern over the current pregnancy. These ranking results correspond to the proportion of time that participants overall divided between the four thematic counseling
categories, as Diagnostic Information and Parental Support were allotted the most amount of time.

It is important to compare the hypothetical approach of GCs in cases of NIPT-DSI to what parents have expressed needing from their providers in other studies in order to ensure that GCs are aligning themselves appropriately with their patients’ needs. One study had found that many parents experience distress during the prenatal evaluation of NIPT-DSI (Finney et al. 2019). Other studies found that parents have concerns over gender identity of their child, gender roles, social stigma from their child’s peers, their child’s future romantic relationships and fertility, and sexual orientation (Crissman et al., 2011). The overall greatest distress for one parent sample surrounded early experiences during the diagnostic period, surgery, and future concerns (Alpern et al., 2016). Although these parental concerns were reported in the context of DSDs diagnosed postnatally, these concerns may bear some similarities to the parental experiences in the situation of NIPT-DSI. While specific potential concerns, such as sexual orientation, gender identity, and social stigmas, were ranked as having lower importance in the present study compared to other discussion topics, participants may have prioritized the more generalized option of providing parental support with the intention of tailoring the discussion of lower ranked topics as they arise in conversation. Despite this, a relatively large proportion of GCs reported some level of discomfort regarding topics that parents have expressed concerns over, including gender identity, gender rearing, surgeries, and sexual orientation. This data suggests that if these topics arise in a genetic counseling session, GCs may not feel comfortable discussing them with the patient. Even if patients do not express concerns immediately in a genetic counseling session, this may be an area in which GCs can provide anticipatory guidance.
When asked what practical steps GCs might take when counseling cases with NIPT-DSI, more than half of participants indicated they would offer some combination of a karyotype, FISH for SRY, and chromosome microarray in both the prenatal and pediatric scenarios. Despite this, other GCs proposed offering a single gene panel and/or exome sequencing. Interestingly, GCs who have experience with more than one prenatal NIPT-DSI case were less likely to indicate that they would offer single gene panels than others in the prenatal scenario ($p = 0.015$). It is unclear why this association exists. Perhaps the outcomes of cases that prenatal GCs with more NIPT-DSI have personally experienced have offered unique insight separate from other influencers of the genetic testing options GCs might offer. Regardless of the reason, this inconsistency in how GCs approach the same clinical scenario suggests that guidelines that clearly specify which testing options to offer patients in cases of NIPT-DSI would be useful to prenatal and pediatric GCs. Of note, the utility and practicality of prenatal testing for DSD has been largely unclear in the past (Adam et al., 2012). Procedures for genetic testing even postnatally still remain ambiguous, as a global DSD update consortium details different first-tier approaches (Lee et al., 2016). One approach consists of stepwise genetic testing beginning with karyotype or FISH for X and Y chromosomes with chromosome microarray, then proceeding to gene sequencing as a last resort; whereas an alternative approach presented in the same consensus statement details exome or genome sequence as a first-tier test (Lee et al., 2016).

Similar inconsistencies appeared regarding which specialist referrals GCs would recommend for their clients with NIPT-DSI. While the majority of participants indicated they would make at least one specialty referral of some kind, there were inconsistencies among participants in the referrals of radiology and surgery, as less than half indicated that they
would make these referrals. Additional referral inconsistencies were seen with urology, where around half of the participants would make the referral and the other half would not. This further supports the potential utility of published guidelines to clarify appropriate referrals in cases of NIPT-DSI. Previously published guidelines have specified that multidisciplinary care of children with DSD should ideally include endocrinology, surgery or urology or both, psychology/psychiatry, gynecology, genetics, neonatology, and, if available, social work, nursing, and medical ethics (Hughes et al., 2006). These guidelines are specified for patients diagnosed with DSD postnatally and, thus, the team of specialists may not be applicable for patients with NIPT-DSI until further evaluation may be performed, particularly if identified prenatally. GCs in this study appear to be attuned to this distinction, as those who encountered NIPT-DSI in a prenatal setting referred to other specialties (psychology or social work, radiology, surgery, and urology) less often than GCs encountering NIPT-DSI in a pediatric setting.

4.3 | Practice Implications

NSGC has published official position statements regarding prenatal cell-free DNA screening and secondary findings on genetic testing, but no specific NSGC position statement exists on the topic of NIPT-DSI. Specialists from various disciplines as well as representatives from support and advocacy groups updated and published a consensus statement regarding the diagnosis and care of individuals with DSDs in 2016 (Lee et al., 2016). This statement comprehensively covers topics including, but not limited to, psychosocial care for patients with DSDs and their families, clinical evaluations, gender assignment, and medical management. Approaches to genetic testing are discussed in these guidelines, although the presented strategy is broad and generalized to DSDs without discussion or consideration of NIPT.
Of note, these guidelines are targeted toward physicians rather than GCs and discuss the role of genetics in a broad sense. Furthermore, less than half of participants in the present study indicated that published guidelines influenced their perceived importance of various topics related to NIPT-DSI, while the majority did indicate previous experience as an influencing factor in both the prenatal and pediatric hypothetical scenario. This indicates that GCs may be relying more on their personal experiences rather than evidence-based guidelines when approaching cases of NIPT-DSI. Having established, evidence-based guidelines has been shown to result in the improvement of the process and structure of clinical care (Lugtenberg et al., 2009). Therefore, given the variability in GC approach to the hypothetical cases of NIPT-DSI and the fact that GCs are encountering these scenarios and caring for these patients in clinic, guidelines should be established for how GCs approach these scenarios. Providing support to GCs in the form of clinical guidelines targeted on the genetic counseling encounter could ensure that patients with the same indication receive the same level of genetic counseling care.

Notably, the percentage of GCs who expressed comfort discussing genital surgeries with patients was low, with only 27% and 29% comfortable in the prenatal and pediatric scenarios, respectively. Based on data gathered in this study, we believe that GCs encounter scenarios of NIPT-DSI in their practice and that they are using their clinical judgment but are generally unsure of how they “should” be approaching these cases. Therefore, genetic counseling training programs and continuing education opportunities should target increasing overall GC knowledge about genital surgeries as they relate to DSDs, in addition to clarifying the GC’s role in these scenarios.

4.4 | Study Strengths and Limitations
This research is the first to investigate GC provider approaches to NIPT-DSI. The sample size was large and representative of GCs as a whole when compared to the 2019 NSGC professional status survey, indicating that these results may be reasonably generalized to other GCs who work in the prenatal or pediatric setting. Additionally, the two scenarios presented in the vignettes were designed to be as similar as possible with exception of the timing of NIPT-DSI being discovered either prenatally or postnatally. This allows for the comparison of participant responses between the two scenarios to be meaningful and appropriate in the context of the study. Additionally, the order in which prenatal or pediatric scenario was presented to participants was randomized to reduce potential bias.

In terms of potential limitations, the survey tool used in this study was unvalidated and, as such, there may be questions that were interpreted differently amongst participants, or not as the investigators originally intended. The hypothetical scenarios used in this study also represent a relatively limited scope of how NIPT-DSI can present in clinical practice. Other situations in which uncertainty surrounding fetal sex occurs may include ambiguous genitalia in conjunction with NIPT fetal sex results and an NIPT results consistent with female sex, but male genitalia on ultrasound or at birth. Because the presented vignettes are just one scenario GCs may encounter, these findings may not apply to all types of NIPT-DSI cases. Finally, selection bias may have skewed collected data if participants with a special interest in NIPT-DSI elected to complete the survey more often than others.

4.5 | Research Recommendations

While this study gathered initial information regarding GC experiences with NIPT-DSI, further evaluation is needed in order to confirm that GCs are approaching real-world cases in the same way that they approached this study’s hypothetical vignettes. This may be achieved
with a retrospective or prospective review of GC decisions and patient outcomes in cases of NIPT-DSI. Additionally, this study provided limited insight into the reasons behind GCs reported genetic testing and specialty referral decisions. Studying why GCs elect various options may help guide efforts to unify the field in the care that they offer patients. In this study, we also attempted to compare topics prioritized by GCs to parental concerns expressed in previous studies. To further evaluate the relationship between genetic counseling care and patient needs, future research may ask families/patients who had genetic counseling for NIPT-DSI if they felt like they received appropriate care and had their concerns addressed. Furthermore, surveying these patients to determine what they find most concerning in relation to the finding of NIPT-DSI will clarify patient needs for these indications.

4.6 | Conclusions

GCs encounter cases of NIPT-DSI in their practice, both in the prenatal and postnatal setting. They are generally equipped to handle these scenarios based on their training in both providing information, along with psychosocial care, however some GCs feel discomfort toward discussion of areas of gender identity, gender of rearing, sexual orientation, and genital surgeries. GCs, in particular, may benefit from further education efforts surrounding genital surgeries as they relate to DSDs due to the high probability of encountering NIPT-DSI in clinic. There is little consensus between GCs regarding tests and referrals they would offer the hypothetical patient. Therefore, guidelines for management of NIPT-DSI targeted toward GCs may alleviate some of the disparity in the theoretical clinical practices of GCs and ensure that patients receive equal and optimum care for the same conditions of NIPT-DSI.
BIBLIOGRAPHY


VITA

Emily Catherine Stiglich was born in Tulsa, Oklahoma, the daughter of Julie Renee Stiglich and Thomas Charles Stiglich. After graduating from Carmel High School, Carmel, Indiana in 2014, she entered Indiana University in Bloomington, Indiana. She received the degree of Bachelor of Science with a major in neuroscience from Indiana University in May 2017. For the next year, she worked as a Registered Behavior Technician in Bloomington, Indiana providing applied behavioral analysis services to children with Autism Spectrum Disorder. In August of 2018, she entered The University of Texas MD Anderson Cancer Center UTHealth Graduate School of Biomedical Sciences.

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