

PRENATAL GENETIC COUNSELORS' PERCEPTIONS OF NON-INVASIVE
PRENATAL TESTING (NIPT): A LOOK AT THE INFORMED CONSENT
PROCESS AND COMMON PATIENT MISCONCEPTIONS

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May 2024

Submitted in partial fulfillment
of the requirements for the degree of
Master of Science in Human Genetics
Sarah Lawrence College

ABSTRACT

Our study investigated prenatal genetic counselors' perspectives on and overall satisfaction with the current practice of informed consent for non-invasive prenatal testing (NIPT). With the routinization of NIPT, it is increasingly necessary for healthcare providers other than genetic counselors to facilitate pretest counseling. This in turn raises concerns that time constraints and non-genetics providers' lack of knowledge about NIPT will leave patients unprepared to make an informed decision about prenatal testing. To explore the frequency of perceived patient misconceptions and their perceived sources and identify potential strategies to address current challenges in the informed consent process, we created an online survey targeting past or currently practicing prenatal genetic counselors consisting of multiple-choice, Likert-scale, and open-ended questions. The survey was distributed to genetic counselors through the National Society of Genetic Counselors listserv as well as directly through professional connections and LinkedIn. Responses were analyzed using descriptive statistics. For open-ended questions, common themes were extracted through inductive analysis. A total of 154 counselors responded and 109 met eligibility criteria. Results showed that OB/GYNs, midwives, and maternal-fetal medicine specialists were most commonly the healthcare providers facilitating informed consent for NIPT. The most frequently reported patient misconceptions were that NIPT screens for all genetic conditions and that NIPT is a diagnostic test, with 82% (n=88) and 78% (n=85) of respondents respectively stating that their patients "sometimes" or "often" hold these beliefs. A majority of respondents expressed feeling frequently dissatisfied with the pretest counseling that their patients had received from non-genetics providers, and they identified a lack of provider education, time constraints, low patient health literacy, and language barriers as potential sources of patient misconceptions. Our results suggest that genetic counselors believe the most common

source of patient misunderstandings about NIPT is non-genetics providers' lack of knowledge about the test. Our results imply that genetic counselors are not confident patients are making fully informed and autonomous decisions when consenting to NIPT. When asked how we might improve the informed consent process for NIPT, respondents were most likely to suggest efforts to standardize non-genetics provider education and to introduce accessible patient resources.

Keywords: NIPT, informed consent, patient misconceptions, barriers

ACKNOWLEDGEMENTS

We would like to thank our incredibly supportive thesis mentors Laura Hercher and Katie Stoll for providing us with their extensive expertise on NIPT practices and research.

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INTRODUCTION

Non-invasive prenatal testing (NIPT) has been in use for over a decade as a screening tool to detect trisomy 13, trisomy 18 and trisomy 21 (known as the “common trisomies”) as well as certain other forms of fetal aneuploidy. Launched in 2011, NIPT was originally described as a diagnostic test and identified as non-invasive prenatal diagnosis (NIPD), which was marketed as a replacement for invasive testing (Norwitz & Levy, 2013). Since that time, NIPT has become best known amongst consumers as a test to identify fetal sex (Agatista et al., 2018; Vanstone et al., 2018). While it has not replaced invasive testing as developers might have hoped, its value as a screening test has advanced the accuracy of prenatal testing beyond the previous standard of care, quad screening. Credit for the technology used by NIPT dates to the discovery of cell-free fetal DNA (cfDNA) in maternal serum in 1997 (Lo, et al., 1997), which enabled the examination of fetal DNA from the placenta as a part of genetic prenatal screening. Since its debut in 2011, NIPT has evolved to include screening for sex chromosome aneuploidies and a number of microdeletion syndromes. In more recent years, commercial applications of NIPT-based tests for single-gene disorders have also been launched (Mohan et al., 2022).

NIPT has become routinized, such that many women may view the screen as “just another blood test” without fully understanding the purpose and possible outcomes of the testing (Cernat et al., 2019). An informed choice is a critical component for prenatal screening programs—including NIPT—with the goal of promoting reproductive autonomy. As it pertains to NIPT, an informed choice is achieved when a woman has a sufficient understanding of the pros and cons of testing to support either a positive or negative attitude towards its use (Kater-Kuipers

et al., 2020). Both accepting and rejecting prenatal screening are expressions of reproductive autonomy.

Professional societies including the American Congress of Obstetricians and Gynecologists (ACOG) and Society for Maternal-Fetal Medicine (SMFM) (ACOG & SMFM committees, 2020), the American College of Medical Genetics and Genomics (ACMG) (Dungan et al., 2023), and the National Society of Genetic Counselors (NSGC) (National Society of Genetic Counselors, 2021) all recommend informing pregnant individuals about all options available during pregnancy, including NIPT, for the purpose of screening for chromosomal abnormalities. These societies agree that informed consent is vital to ensure that patients are aware of the risks, benefits, and limitations of each testing option, as well as that patients are making an informed choice when electing to proceed with NIPT. Routinization of NIPT use has the potential to lead to uninformed decision-making which in turn may leave pregnant individuals emotionally unprepared and vulnerable when they receive results that can be disturbing or confusing (Mozersky, 2015; Michie & Allyse, 2015). Kater-Kuipers et al. (2020) state that “women might routinely accept NIPT as a screening test for trisomy 21, 18 and 13 and may not be prepared for abnormal test results.” The concept of a “screening trap” has been described, where patients are placed on a pathway to invasive follow-up and potential termination of the pregnancy without fully understanding the consequences prior to the screen (de Jong & de Wert, 2015).

Inadequate informed consent regarding the accuracy of NIPT and the potential for false positive results can lead to poor outcomes for parents and families. Families should be aware that NIPT is not diagnostic and that both false negative and false positive results can occur for many reasons (Zhang et al., 2015). In fact, the positive predictive value of the test can be as low as

51%-73% on average for trisomy 21 for a variety of patient demographics, and increasingly lower for less common positive reports (Xiang et al., 2023). A 2015 case report described a couple who relied on and were reassured by their low-risk NIPT result even after a prenatal cardiac defect was seen on ultrasound who then gave birth to a child with trisomy 21 (Smith et al., 2015). More complex cases reveal instances of fetoplacental mosaicism for trisomy 21 found after low-risk NIPT, discovered through cytogenetic analysis following termination of pregnancy due to complex ultrasound findings (Feresin et al., 2022). With regard to these false negatives, accurate and complete pre- and post-test counseling for NIPT is imperative for families to understand the residual risk for aneuploidy, the importance of sonography, and the availability of confirmatory diagnostic testing.

False positives are also an issue. Two separate case reports from 2014 describe false positive abnormal results. In one, a patient's NIPT results were positive for trisomy 13; subsequent ultrasound examination showed no anomalies and amniocentesis confirmed a normal 46,XY fetus (Verweij et al., 2014). In the other report, a patient's NIPT results indicated a female fetus with 99% risk for Turner syndrome (monosomy X); invasive diagnostic testing was later done to investigate the fetal karyotype, revealing discordant results. Chorionic villus sampling (CVS) results indicated a mosaic male karyotype (46,X,idic(Y)/45,X), and amniocentesis confirmed a male karyotype with an isodicentric Y chromosome and no signs of mosaic monosomy X. The patient in question had expressed intent to terminate the pregnancy based on the NIPT and CVS results; it wasn't until an amniocentesis suggested that the fetus would likely only suffer from infertility that she chose to proceed with the pregnancy (Srebniak et al., 2014). If the potential for NIPT to produce false positives is not well understood by

patients and providers, there could be an increase in the number of healthy fetuses terminated (Allyse et al., 2015).

Given the recommendations that NIPT should be offered to all pregnant individuals, the question arises: is NIPT being discussed appropriately with patients in the clinical setting? While the NSGC's position statement from 2021 recommends that NIPT be offered to all patients in the context of other available screening and diagnostic testing options, and that pretest counseling should include discussions on patient values and needs as well as the benefits and limitations of NIPT, access to competent pretest counseling can be difficult to obtain due to a shortage of certified prenatal genetic counselors (National Society of Genetic Counselors, 2021; Dickens, 2014; Allyse et al., 2015). With the integration of NIPT into the routine care of pregnant patients, pretest counseling by a genetic counselor for every patient has become unworkable as a model, both from a personnel and a cost perspective. Inevitably, the healthcare providers most often discussing NIPT with patients are obstetricians, midwives, and primary care providers (Brewer et al., 2017), and most pregnant patients now only meet with a prenatal genetic counselor when NIPT identifies an increased risk of a potential genetic condition (Stoll & Jackson, 2020). Studies have indicated that healthcare providers who do not specialize in genetics lack the knowledge and time during appointments to discuss testing options and provide genetic services to patients (Chan et al., 2018; Gammon et al., 2016; Swaney et al., 2015).

It was reported in 2001 that, during medical school, physicians spent under thirty hours training in genetic concepts; when these measures were reassessed in 2013, there was no substantial increase in the training time (Greendale & Pyeritz, 2001; Benseid et al., 2014). Since the introduction of NIPT in 2011, one would think that this training time would have increased, particularly as genetic testing has become a part of nearly every medical specialty; however,

further studies have only continued to show that medical schools have not sufficiently trained physicians in genetics to a level where they feel comfortable discussing it in clinical practice. Haspel et al. (2021) asked postgraduate residents about their training in genetics and interactions with genetics professionals. Over 30% of respondents reported that they had not had any training in genetics and genomic medicine during medical school; just over half reported interacting with a medical geneticist during their training, while 32% had interacted with a genetic counselor. When asked to self-assess their understanding of genetics, 32% rated their knowledge of genetics-related topics as good or better than good (Haspel et al., 2021). These findings show that, even as genetic testing has become more and more mainstream in our healthcare system, the training that physicians receive has not increased to meet that integration.

Previous studies have aimed to assess how non-genetics providers acquire knowledge about NIPT outside of medical school training. Most providers reported engaging in didactic education or seeking out medical literature to learn more about NIPT, though commercial laboratories were also often cited as a common source of information (Farrell et al., 2016; Swaney et al., 2015). Since the introduction of NIPT in 2011, studies have been conducted that assess providers' technical knowledge of it as a screening tool. Researchers found that, while most responded correctly, there was evidence of fundamental misunderstandings, with Haymon et al. (2014) and Brewer et al. (2016) each reporting that upwards of 15% of respondents considered NIPT to be diagnostic for fetal aneuploidy. Additionally, a more recent study found that 19.4% of general practitioners agreed that they would offer termination of pregnancy immediately following a positive NIPT result (Chan et al., 2018). This lack of understanding on providers' parts has not gone unnoticed by patients, with some reporting a dissatisfaction with their pretest counseling experience, stating that they felt their clinicians lacked knowledge (Cain

et al., 2019) and were not sufficiently informed about the technology (Floyd & Allyse, 2016; Vanstone et al., 2018; Lau et al., 2016).

A second consideration is that non-genetic providers often have a bevy of other topics to discuss with patients and limited time allotted for each appointment, essentially requiring that any discussion of NIPT be brief. Cain et al. (2019) found that non-genetic providers do not always spend adequate amounts of time discussing prenatal aneuploidy testing with patients. While just under 50% of patients reported that their providers spent anywhere from five to fifteen minutes on this discussion, just over a third of patients reported that the discussion was less than five minutes (with some reporting that there was no conversation at all). Similarly, Martin et al. (2018) found that providers on average spent nine minutes discussing prenatal testing, even when allocated thirty minutes for that purpose.

Providers themselves have voiced concerns about the limited time available to properly counsel patients about their prenatal testing options (Farrell et al., 2015; Gammon et al., 2016). As prenatal testing continues to expand and the number of conditions on screening panels increases, providers are often left with little guidance on the level of detail their patients prefer and may have less experience in helping them choose between different screening and testing options (Allyse et al., 2017). Patients have also acknowledged the impact of limited time during appointments, noting that they agreed that consultations were often too short for adequate counseling about NIPT (Lau et al., 2016).

Additionally, research suggests a discrepancy between what providers may prioritize when facilitating informed consent for NIPT and the information patients would most value having about NIPT. When providers were surveyed about the topics they considered most important to cover, respondents ranked the clinical utility of the test highest, followed by risk to

fetus or mother and test sensitivity, while the range of conditions covered by NIPT was ranked near the bottom (Sayres et al., 2011). Similarly, Hill et al. (2016), found that providers most often emphasized test accuracy and timing. In the same study, Hill et al. (2016) found that pregnant patients were themselves most concerned with test safety and comprehensive information on both the test and conditions covered. Farrell et al. (2014), found that most patients prioritized wanting to understand the implications of NIPT—including which genetic conditions it could and could not identify—and details on the quality of life and expected features associated with these conditions. Similar results were found by Dane et al. (2018), who reported that patients felt it was important for them to know what conditions NIPT can and cannot test for, as well as what the pros and cons of NIPT are compared to other prenatal tests.

Today, prenatal genetic counselors are most often seeing patients post-NIPT, when healthcare providers are referring patients with positive test results indicating an increased risk for a condition (Stoll & Jackson, 2020). As such, they both see and deal with the aftereffects of any inadequacies in the informed consent process. While evidence-based recommendations show the value of a comprehensive pretest informed consent process, even before NIPT was introduced, there were concerns that healthcare providers would not see consenting for prenatal genetic testing as requiring the same stringent standards as consenting for invasive procedures (de Jong et al., 2010). A study from 2014 asked genetic counselors about their views and experiences with cell-free fetal DNA testing as it was first being implemented in a clinical setting. When asked whether pretest counseling was necessary for cfDNA testing, 98% of respondents chose some gradation of agree; however, when asked whether this pretest counseling could be administered equally well by a health professional other than a genetic counselor, 63.1% of respondents chose some gradation of disagreement (Horsting et al., 2014).

Building on Horsting et al., we chose to survey prenatal genetic counselors to get an updated view on the practice of informed consent for NIPT and to assess the implications of current practice for patients in a post-test session.

METHODS

Participants

Certified genetic counselors with experience in prenatal genetic counseling involving pre- and/or post-test counseling for NIPT were recruited for this study. Both past and current prenatal genetic counselors were eligible.

In an effort to reduce the possibility that bots would complete our survey, we began the survey with a mandatory two-part question that asked participants to write in their favorite television show and character. Of the 154 survey responses, forty-eight submissions were flagged and forty-five were excluded from data analysis as potential bots or duplicate entries from a single participant. We used the following rationale to determine exclusions. To start, we excluded participants that did not include both a television show *and* a character for the initial two-part bot-identifying question. One response provided neither a television show nor a character and was thus flagged, but as the respondent noted that they did not watch television and instead provided their favorite book and book character, we ultimately decided to include this entry. Responses with duplicate answers (same show and different character or same show and same character) submitted in close succession to each other were flagged. For these submissions, we looked at the survey start time to determine if they seemed legitimate; if the surveys were started around the same time (within ninety seconds of each other), we counted *all* as submitted by a bot and they were thus excluded. There were four instances of this in total.

Two additional surveys listed the same television show and character, but the start and end times of these submissions were not close together and the respondents in each case provided credible emails associated with known academic institutions for the raffle; we considered this adequate reassurance that these submissions were not from bots, and they were included in data analysis.

After this first filter, we looked again for bots by examining the emails provided for the gift card raffle and flagged any entries that had duplicate emails. For the six entries with duplicate emails (three pairs of responses), we reviewed the start times of the survey as well as the responses to the survey questions. If the submissions were started around the same time (within ninety seconds of each other), we considered them to be from a bot. If the submissions were started at different times and the responses to multiple survey questions were different, we also considered them to be from a bot. In both of these instances, the submissions were excluded from our final data analysis. If the submissions were started at different times but the responses to questions were the same, we decided to include the earlier submission in our final data analysis but exclude the later submission.

Instrumentation

An online survey was constructed for data collection. Survey questions were formulated in consultation with relevant literature, including prior studies that assessed perspectives on the informed consent process for NIPT (Dane et al., 2018; Farrell et al., 2016). The survey consisted of fourteen questions, excluding the initial two-part question and final email collection question. Of these fourteen, there were eleven multiple-choice questions, two open-ended response questions, and one Likert-scale question that consisted of four sub-questions on a 5-point scale ranging from “never” to “always.” Demographic questions in the survey asked about participant age, percentage of practice spent doing prenatal genetic counseling, years of experience

providing prenatal counseling, and employment setting. We asked participants about who consented their patients for NIPT, what they felt were the most important aspects of the NIPT informed consent process, and what they felt were barriers to best practices in NIPT consenting. Additionally, we asked participants to rank how often they felt patients misunderstood NIPT as: a test for all genetic conditions; a test solely for fetal sex; a test solely for trisomy 21 (Down syndrome); or a diagnostic test.

Procedures

Participants were recruited via the online survey link distributed by the NSGC Student Research Survey Program to a members' listserv on December 6, 2023. A follow-up posting was sent on January 10, 2024. Additionally, a recruitment poster was created including the study's goals, survey link, and our contact information; this was shared on LinkedIn and distributed to current and previous clinical rotation supervisors and colleagues for additional recruitment. The survey was closed on January 31st, 2024.

Upon opening the survey, participants were presented with a consent form which stated our research question, our goals, how we would use participants' information, and that participants could withdraw at any point during the survey. Participants were informed that by completing the survey and providing us with their email address, they would be entered into a raffle to win one of ten \$25 Amazon gift cards for their participation in the study. Email addresses were separated from participant responses after closing the survey. Study funds were provided by Sarah Lawrence College. The online survey platform SurveyMonkey was used to collect survey responses. De-identified data was stored in a password-protected file. This study was approved by the Sarah Lawrence College Institutional Review Board on November 26, 2023.

Data Analysis

Excluding open-ended and demographic questions, all questions were measured as categorical outcomes or on a Likert-scale. Descriptive statistics for qualitative multiple-choice survey questions were calculated using SurveyMonkey's statistical software and Microsoft Excel. For open-ended responses, responses were analyzed and common themes were extracted and labeled in Excel. Responses were quantified in percentages after coding.

RESULTS

Demographics

A total of 109 survey responses were eligible for the study. Participants were asked their age, percentage of practice spent providing prenatal genetic counseling, years of experience providing prenatal counseling, and employment setting. The response rate was 100% for all demographic questions. Approximately half of all respondents were under the age of 30 (49.5%, n=54), 31.19% (n=34) were aged 30-39, 12.8% (n=14) were aged 40-49, 5.5% (n=6) were aged 50-59, and 0.9% (n=1) were over 60 years old. The majority of respondents reported that more than half of their practice consisted of prenatal genetic counseling (76.1%, n=83). Just over half of participants had provided prenatal genetic counseling for between 1 and 5 years (50.5%, n=55). The majority of respondents were employed by a public hospital or clinic (65.1%, n=74), with fewer working in private practice (21.1%, n=23), and laboratory positions (8.3%, n=9). Full demographics are reported in Appendix A.

NIPT pre-test providers

Participants were asked to describe who primarily provides their patients or clients with pretest counseling for NIPT (Figure 1). Participants were asked to select all that apply from a list

of ten options, with space provided for additional answers if needed. From this list, participants selected obstetricians and gynecologists (OB/GYNs) most frequently (85.2%; n=92). Second most commonly chosen, in a tie, were midwives and maternal fetal medicine (MFM) specialists (50% of all participants; n=54 and n=54, respectively). Less than half of participants indicated that pre-test counseling was completed by another genetic counselor (46.3%, n=50). Less than a third of participants indicated that primary care providers (i.e. MDs, nurse practitioners) or other clinical staff were facilitating this process (31.5%, n=34 and 24%, n=26, respectively). While not a provider, “informational resources” was offered as an option; eight participants (7.4%, n=8) selected this as a source for pretest counseling. Five respondents (4.6%, n=5) were unsure who was providing pretest counseling for their patients and another four (3.7%, n=4) reported that “no one” was providing pretest counseling. Notably, one respondent (0.9%, n=1) shared that “genetic counseling assistants meet with all patients having NIPT drawn at our hospital.”

Patient Misconceptions

Participants were asked four questions about how frequently patients exhibit the following misconceptions: (1) that NIPT screens for any genetic condition, (2) that NIPT only screens for fetal sex, (3) that NIPT only screens for Down syndrome, and (4) that NIPT is a diagnostic test. A Likert-scale format was used for these questions, with possible responses ranging from “never” to “always.” Complete results can be seen in Figure 2. The majority of respondents reported that their patients “sometimes” (52.8%, n=57) or often (28.7%, n=31) believe that NIPT screens for any genetic condition. Nearly two-thirds of respondents said that their patients “sometimes” (33%, n=36) or “often” (31.2%, n=35) believe that NIPT only identifies fetal sex. Most respondents said that patients “sometimes” (39.4%, n=43) or “often” (29.4%, n=32) believe that NIPT only screens for Down syndrome. Finally, most respondents

said that their patients either “sometimes” (41.3%, n=45) or “often” (36.7%, n=40) believe that NIPT is a diagnostic test, while 20.2% of participants (n=22) said that patients “rarely” believe that NIPT is a diagnostic test.

Participants were asked to select any or all factors that they felt contributed to patient misconceptions about NIPT, given the following list: limited time in pre-test sessions, language barriers, misinformation from NIPT media/marketing, physicians’ limited familiarity and experience with NIPT, limited staff and resources to assist with counseling, patients’ health literacy, and other (Figure 3). The most commonly selected contributors to patient misconceptions were physicians’ limited familiarity and experience with NIPT (77%, n=84), patient’s health literacy (72.5%, n=79), limited time in pretest sessions (66%, n=72), misinformation from NIPT media and marketing (56%, n=61), limited staff and resources (55%, n=60), and language barriers (48.6%, n=53). Respondents also had the option to write in other factors they had encountered, and one participant reported that “confusing reports for certain findings like atypical findings and high risk due to low fetal fraction” was a contributor to patient misconceptions.

Participants were then asked to select what they felt was the single most important factor contributing to patient misconceptions about NIPT (Figure 4). Participants most frequently described physicians’ limited familiarity and experience with NIPT (33.9%, n=37), limited time in pre-test sessions (24.8%, n=27), and limited staff and resources to assist with NIPT counseling (13.8%, n=15) as the most important source of patient misconceptions. Of the six (5.5%, n=6) participants who selected “other” and wrote in the text box provided, one reported that the most important contributor to patient misconceptions was “the lack of any pretest counseling at all by the OB/GYNs.”

Barriers to Informed Consent

Participants were asked which of the following they felt would help reduce patient misconceptions: (1) printed educational materials about NIPT, (2) electronic resources, (3) greater access to genetic counselors, (4) shared medical appointments (where genetic counselors see patients concurrently with other healthcare providers), (5) group information sessions for patients, and (6) “other”. Participants could select more than one response. Over two-thirds of respondents selected greater access to genetic counselors (68.8%, n=75). The majority of respondents selected printed educational materials about NIPT (55%, n=60) and shared medical appointments (53.2%, n=58). Among those participants who selected “other” (10%, n=11), responses included: “better and increased OB/GYN education for NIPT,” “eliminating the term ‘gender’ from counseling,” and “more genetic counselors present on social media platforms to explain NIPT.” Full responses are reported in Figure 5.

Participants were asked to indicate the most important components of informed consent for NIPT; multiple options could be selected. Nearly all respondents reported that discussing the “benefits and limitations of NIPT” was the most important component of the informed consent process (91.8%, n=100). The majority of respondents also indicated “clarifying conditions included in NIPT” (82.6%, n=90), “the option to decline all screening/testing” (69.7%, n=76), and “discussing the patient’s values, preferences and needs” (56.9%; n=62). Less than half of respondents selected “no irreversible actions should be taken based on NIPT,” important “testing details,” or “the option to pursue diagnostic testing as a first-line approach.” Six participants who chose “other” (5.5%, n=6) mentioned “methodology for return of results,” “possibility of no results,” “emphasis that NIPT is a screening tool,” and “clarification that NIPT is not a gender test.” Complete results can be seen in Figure 6.

Satisfaction and modifications for current informed current practice

At the end of the survey, respondents were asked two open-ended questions. For the first, respondents were asked if they were satisfied with the current informed consent process for NIPT in the pretest setting and for the second if there were any modifications they would wish to make. In total, eighty-four participants (77.1%, n=84) responded to this question. Twenty-eight respondents (33.3%, n=28) wrote that they were satisfied with the informed consent process while forty-nine (58.3%, n=49) wrote that they were not satisfied; six participants (7.1%, n=6) wrote in a response but did not clarify if they were or were not satisfied.

Fifty-four out of the eighty-four (64.3%, n=54) responses suggested modifications to their current informed consent process. Figure 7 displays eleven themes for proposed modifications to the informed consent process. The most commonly seen theme was a request for changes involving external providers, including increasing provider education, improved follow-up, and more emphasis placed on test outcomes (40%, n=26). Other common themes included more genetic counselor involvement (i.e. more genetic counselors doing pretest counseling) and additional resources in either paper or video form. Of note, two respondents (3.1%, n=2) wrote that they wished there was “more” or “any” pretest counseling. Full data of coded responses can be seen in Figure 7.

DISCUSSION

This study investigated prenatal genetic counselors’ perspectives on current NIPT pretest counseling practices. It was hypothesized that other healthcare providers such as OBGYNs and other PCPs, rather than genetic counselors, would be the primary facilitators of the informed consent process, as per previous studies (Brewer et al., 2017, Haymon et al., 2014; Allyse et al.,

2015; Horsting et al., 2014; Johnston et al., 2017). We asked prenatal genetic counselors, who commonly see patients for post-test counseling, about the common misconceptions their patients have regarding the clinical utility of NIPT, and to what they attribute these misconceptions. Furthermore, we asked participants if they were satisfied with the current NIPT informed consent process in their practice and what modifications they might suggest to improve the accuracy and completeness of patient understanding.

A total of 109 participants were eligible for study inclusion. The majority of participants shared that more than half of their practice consists of prenatal genetic counseling and that they are employed by a public hospital or clinic. Just over half of participants had provided prenatal genetic counseling for between 1 and 5 years (50.5%, n=55). The 2023 NSGC Professional Status Survey (PSS) found that the majority (83.8%) of prenatal genetic counselors' primary area of practice is prenatal genetics (NSGC, 2023). Similarly, the PSS reports that most (30%) prenatal genetic counselors have between 1 and 4 years of counseling experience. In these respects, our study cohort, though small, accurately represents our target population.

Informed Consent

The ethical basis of prenatal screening is informed decision making, as evidenced by informed consent. A successful informed consent process provides patients the opportunity for genuinely autonomous decisions about whether or not to opt in to prenatal screening (Bunnik et al., 2013; Walker, 2013). The current NSGC position statement on prenatal cell-free DNA (cfDNA) screening outlines the components that should be offered during pretest counseling to ensure informed consent: (1) option of pursuing diagnostic testing as a first line approach, (2) option to decline all screening/testing, (3) discussion of the individual patient's values, preferences, and needs, and (4) benefits and limitations of cfDNA screening. Our participants

generally agreed on these as the essential components of a valid informed consent process but prioritized some points of discussion more than others.

In our study, “discussion of the benefits and limitations of NIPT” was the most commonly identified as an important component of the informed consent process. The second most frequently identified component was “clarifying which conditions are included in the screening.” Notably, participants were permitted to select multiple components in their response, allowing them to emphasize all parts of the session they considered most important. In our cohort, the majority of prenatal genetic counselors are providing pretest counseling “some of the time.” A smaller proportion of participants provide pretest counseling “most of the time,” and no participants claimed they “always” provide pretest counseling. Our results do show adherence to the majority of the current NSGC guidelines for pretest counseling; the component least likely to be identified as important was “the option to pursue diagnostic testing as a first line approach.” Possibly, that this component was selected less frequently by respondents may reflect the fact that patients rarely seek this option.

Genetic counselors are the providers most likely to be trained to adhere to recommended counseling frameworks to allow for autonomous decision making. In contrast, other professional societies and the literature in general do not provide much guidance on a framework for pretest counseling that other providers might utilize in a clinical setting. But previous studies have shown that most genetic counselors are only meeting with pregnant patients upon detection of a high-risk result (Stoll & Jackson et al., 2020), and a prior survey of obstetricians found that only 30% referred patients with positive NIPT results to a genetic counselor (Jelin et al., 2016). These results suggest that non-genetics providers may be the only source of information for many patients, even those considered to be high-risk. While the consequences of inadequate

informed consent may seem minor for those with low-risk pregnancies—as many of these patients will have a negative test with no subsequent follow-up—they are not minor for those who test positive, who may feel blindsided by information on conditions they hadn't previously been aware of or taken into consideration before testing.

Patient Misconceptions

Patient understanding of NIPT and other prenatal screening options has been demonstrated to be inadequate in prior studies (Piechan et al., 2016; Farrell et al., 2015; Gourounti & Sandall, 2008; Rowe et al., 2006). Patient understanding was a focal theme of our study as we wanted to investigate how frequently genetic counselors encounter patient misconceptions today. Our results show that genetic counselors find that their patients “sometimes” or “often” believe that NIPT screens for all genetic conditions, only identifies fetal sex, only screens for Down syndrome, and is a diagnostic test. Few participants reported that patients “never” agree with these common misconceptions. While we did not assess patient understanding directly, prenatal genetic counselors' views remain a valuable window into the patient experience and their observations suggest a need for further investigation into the root causes of patient misconceptions and proposed remedies for patient knowledge gaps.

Sources of Misconceptions

With NIPT more integrated into standard-of-care prenatal practice, there are not enough certified prenatal genetic counselors to meet the demand of pretest counseling for every patient. As such, this task necessarily falls to other providers. Our study found that OB/GYNs, midwives, and MFM specialists were the providers most often facilitating pretest counseling for NIPT, and this finding is in line with previous studies (Haymon et al., 2014; Allyse et al., 2015; Horsting et al., 2014; Johnston et al., 2017). Previous research has shown that these providers spend limited

time discussing NIPT and may fail to address important components of informed consent as outlined by NSGC's most recent position statement. Similarly, genetic counselors in our study report that their patients were often inadequately informed. In response to an open-ended question about NIPT experiences, numerous participants reported frustrations with what other providers are sharing with patients regarding the purpose of NIPT and the misinformation that occurs as a result. Participants wrote: "many healthcare providers don't understand NIPT," and "I'd really like to see the referring providers have some better education for themselves that they can pass onto their patients... many simply do not understand the testing or its implications." One wrote that their patients have been told that "[NIPT] 'works like magic' to find fetal Down syndrome and other anomalies." Another wrote that they "wish[ed] OB/GYNs would do a better job of describing NIPT," with others adding that their patients are being told that NIPT "replaces diagnostic testing," "is a diagnostic test" itself, or "only screens for fetal sex."

Many of our participants expressed a wish that providers would, at minimum, emphasize that the purpose of NIPT is to inform fetal risk of chromosome aneuploidies, including sex chromosome aneuploidy. If providers aren't emphasizing or even discussing the range of possible positive results, it can lead to patient confusion and/or decisional regret. One participant wrote, "I have seen an increase in families who receive sex chromosome aneuploidy results who tell me this possibility was not addressed by their provider prior to screening. Oftentimes...they feel this is information they may not have wanted." Similarly, another wrote that many of their patients with atypical NIPT findings have expressed that they "wish they would have known of the possible results and may have never pursued NIPT had they been properly informed."

While the majority of respondents place the onus of misinformation on other healthcare providers, there were some who thought it could instead be attributable simply to information

overload, acknowledging that there is an abundance of information shared in a first appointment, “so even with thorough counseling and checking for understanding, not everything may stick by the time the results are available.” As in prior studies, our participants acknowledged that there is “so much to go through at initial prenatal appointments, [making] it hard to discuss all testing and screening options in detail.”

Numerous participants suggested that genetic counselors could create educational materials for non-genetic providers to standardize NIPT pretest counseling or hold educational meetings with referring OB offices, echoing calls for professional collaborations and educational materials from a 2017 panel (Allyse et al. 2017). Indeed, such training resources have been offered to providers in the United Kingdom and have resulted in a statistically significant increase in providers’ confidence discussing NIPT with patients as well as both perceived and actual knowledge about NIPT (Oxenford et al., 2017). This resource was in the form of a forty-minute training session, which illustrates that these positive changes are feasible without demanding too much time from providers’ already-busy schedules.

Limitations

Our survey was distributed through the NSGC Listserv which is only available to NSGC members. While there was an attempt to recruit in alternative ways including LinkedIn and snowball sampling through professional connections, future studies could utilize professional conferences or alumni networks to recruit non-members. The survey was only accessible in English, which might have been a barrier to some potential survey respondents. A notable limitation of this study is the potential inclusion of responses from bots searching for financial compensation through survey participation. While we attempted to be diligent in removing them,

it is possible that undetected bots may have minimally skewed our results. For future data collection, a more reliable method of distinguishing human and bot responses would be valuable.

In retrospect, it was noted that prenatal genetic counselors expressed concerns in open-ended questions about patient misconceptions not anticipated in the study design (Figure 2). A more inclusive approach, including adding “other” as a multiple-choice option, could have opened discussion for a practice-based perspective that went beyond the list of patient misconceptions already established in the literature. In a similar way, the study did not investigate genetic counselors' perspectives on the competency of specific healthcare providers' pretest counseling. This analysis was not within our study aims, however in retrospect, it could have further stratified our data on provider education. Lastly, survey data were analyzed using qualitative methods which included subjective coding and non-statistical findings, thus minimizing the generalizability of our results.

Further Research Recommendations

In our study, we asked participants about which providers facilitate pretest counseling for NIPT and counselors' overall satisfaction with that process, but we did not ask them if there was any difference in that satisfaction based on what type of provider their patients had previously seen. To our knowledge, there has not been a study that looks specifically at how pretest counseling may differ when facilitated by an OB/GYN, midwife, MFM specialist, or other PCP, nor at how patients' knowledge scores may vary based on the facilitating provider. Looking at these differences may suggest that different providers focus on different aspects of NIPT, which could further highlight where gaps in care occur and where improvements can be made.

Numerous participants suggested a means of standardizing the pretest and informed consent process so that all patients are receiving the same information, regardless of the type of healthcare provider they are seeing. Future studies could collaborate with a panel of prenatal genetic counselors and other genetic specialists to determine how best to standardize this process in a time-efficient manner and how to assess its effectiveness in improving on patient knowledge of NIPT.

When asked about additional experiences with NIPT that they wanted to share, one participant wrote the following: “[I] am in a state where patients have the privilege to wait for ultrasound and genetic testing results before making termination decisions, which allows me to feel more comfortable with letting post-test counseling for positive results make up for any lack of understanding in pretest.” Since the overturning of *Roe v Wade* in 2022, there has been research conducted on the implications for prenatal testing (Raymond et al., 2023) which suggests that some patients may opt out of screening completely move directly to diagnostic testing to meet gestational age-based abortion timelines. For states where abortion laws are restricted, further studies could be conducted to assess the impact of abortion restrictions on NIPT practices, including NIPT uptake, how providers frame the benefits and limitations of NIPT, and how sequential diagnostic testing is discussed.

CONCLUSION

Since its introduction in 2011, NIPT has become routinized in the care of pregnant patients regardless of their risk status. As a result, providers other than genetic counselors have increasingly become facilitators of pretest counseling for NIPT informed consent for these patients. Given the time constraints and lack of robust genetics education that these providers

may have, concerns have been raised about the quality of this counseling and whether patients are making informed choices. Our data showed that prenatal genetic counselors were often dissatisfied with the pretest counseling that their patients received from non-genetic providers like OB/GYNs, midwives, and MFM specialists, and most often believed this to be due to physicians' limited familiarity and experience with NIPT as a screening tool. Participants reported that greater access to genetic counselors, access to educational resources, and standardized physician education could be worthwhile means to help address this issue and lessen patient misconceptions about NIPT that they encounter in post-test sessions. Future research can work towards distinguishing the efficacy of NIPT pretest counseling as provided by various healthcare professionals and improving physician knowledge of the test. Efforts to standardize the informed consent process could help mitigate physician time restrictions.

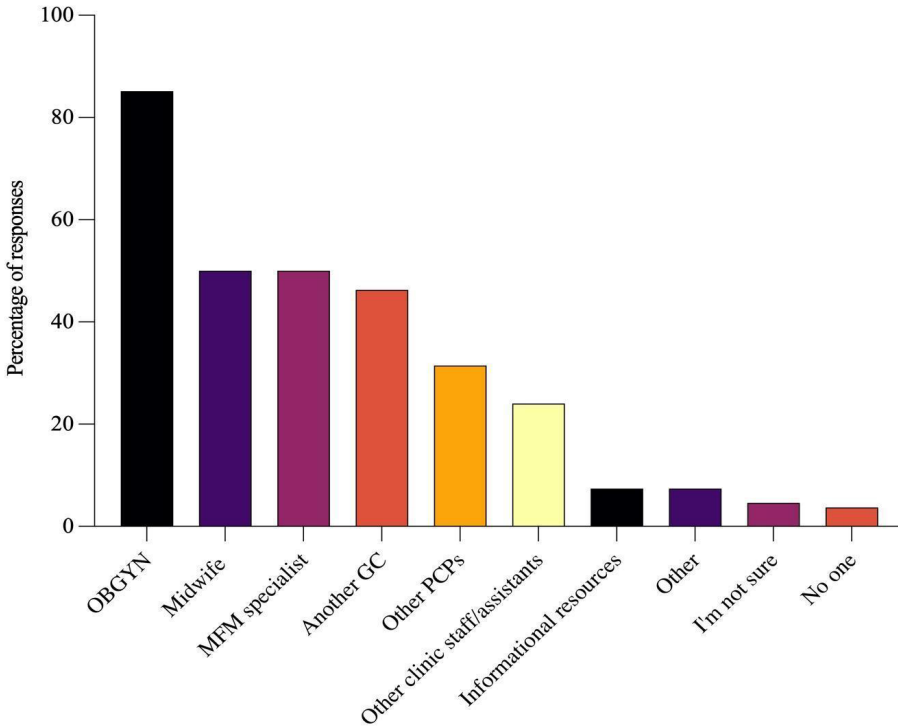


Figure 1. Healthcare providers who provide pretest counseling for NIPT as indicated by study participants (n=109). OB/GYN = obstetrician/gynecologist; MFM = Maternal Fetal Medicine; GC = genetic counselor; PCP = primary care provider.

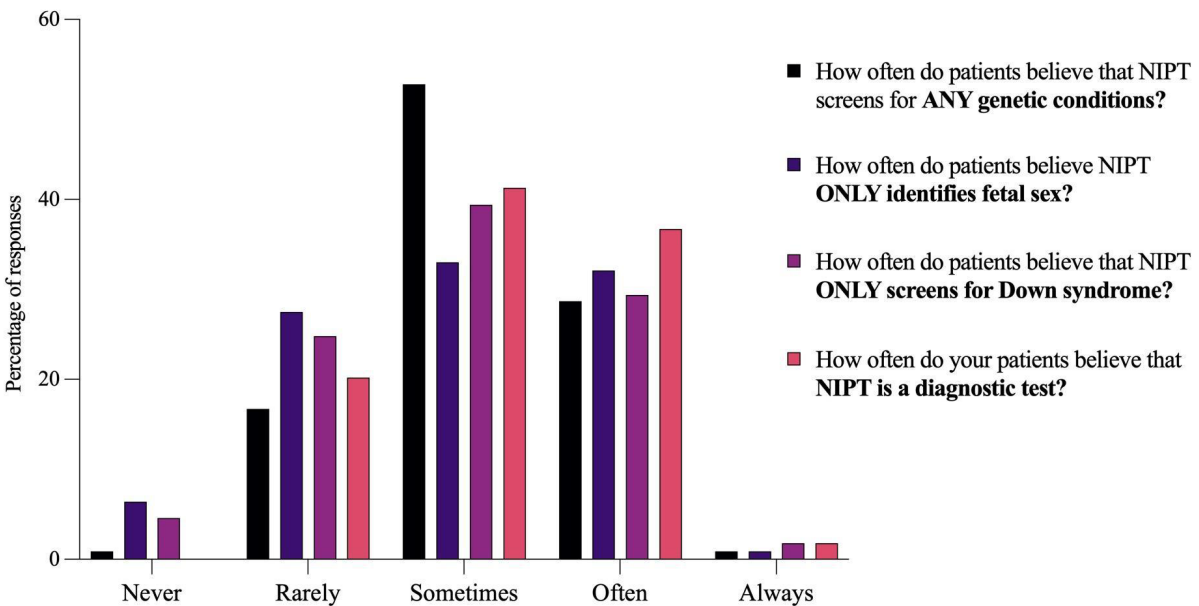


Figure 2. Prenatal genetic counselors' assessment of patient misconceptions about NIPT (n=109). Frequency for each of four possible misconceptions was ranked on a 5-point Likert scale.

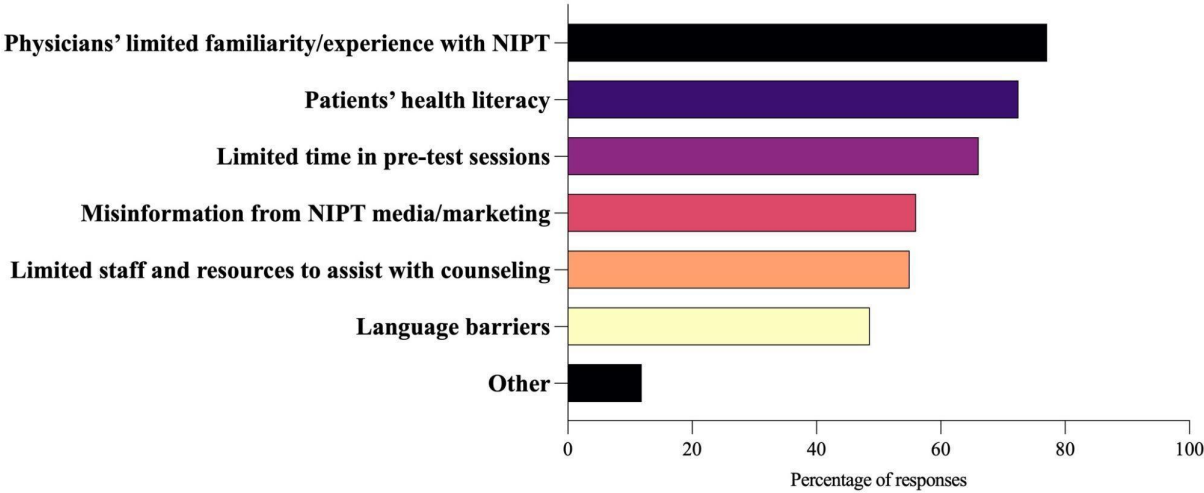


Figure 3. Participants' (n=109) views on factors contributing to patient misconceptions about NIPT. Participants could select multiple responses.

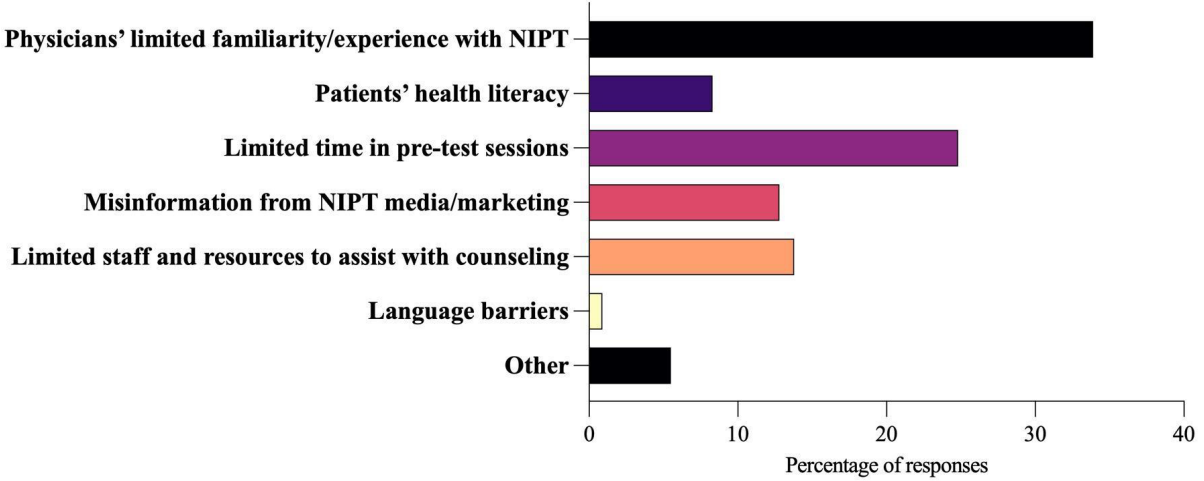


Figure 4. Most important factor contributing to patient misconceptions about NIPT. Responses are presented as percentages (n=109). Participants could only select one response.

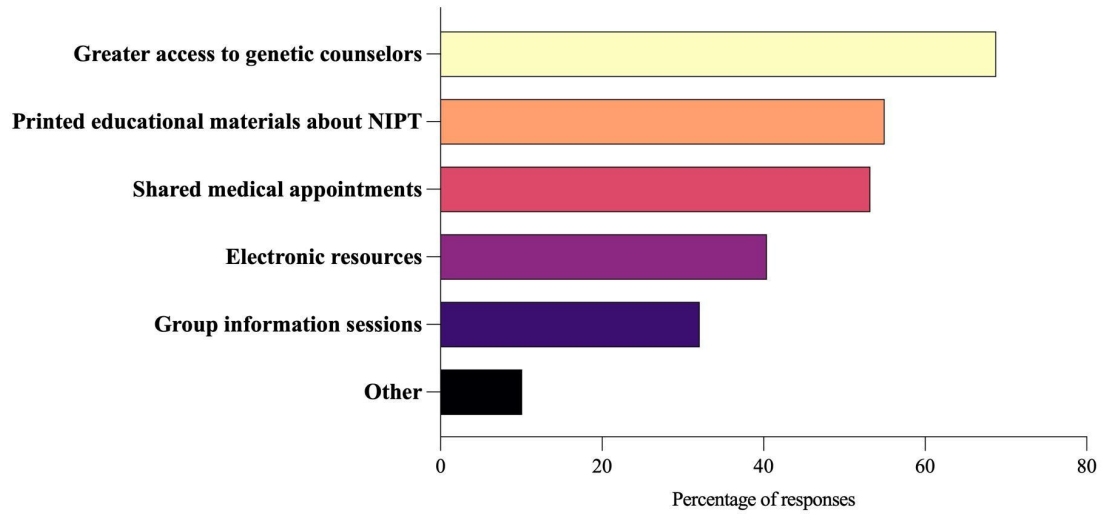


Figure 5. Participants' (n=109) views surrounding factors to overcome patient misconceptions about NIPT. Responses presented in percentages. Participants could select multiple responses.

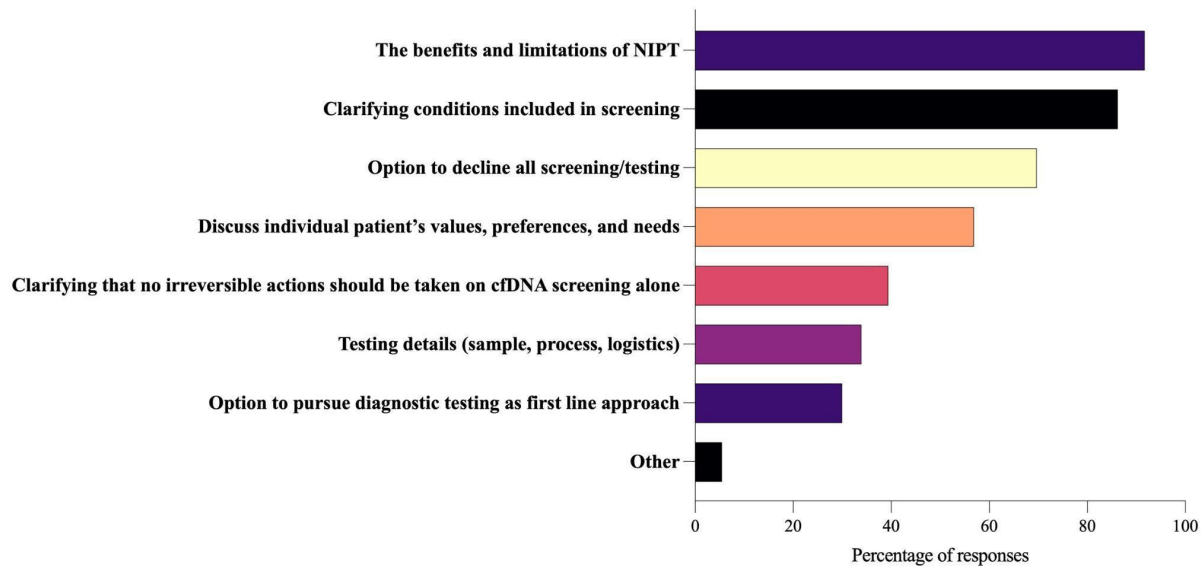


Figure 6. Participants' views on the most important components of the informed consent process for NIPT as percentages (n=109). Participants could only select one response.

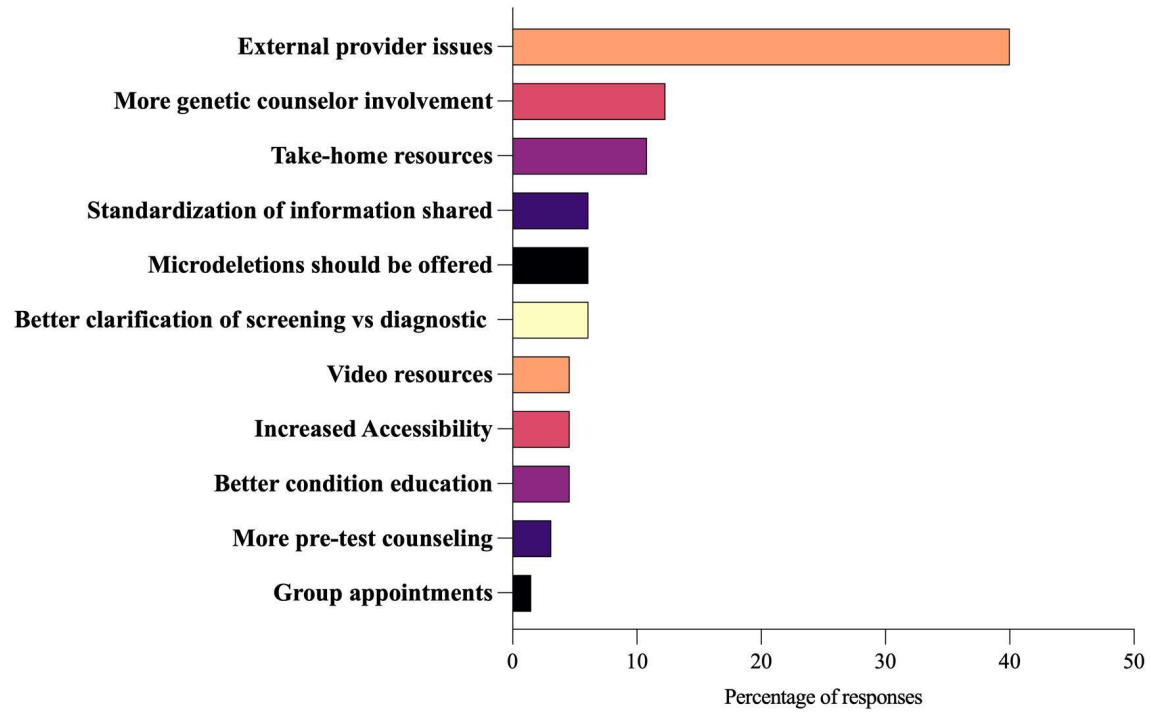


Figure 7. Proposed modifications to improve current NIPT informed consent process. There were 54 participants who provided responses; some responses proposed multiple codable modifications (n=65).

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APPENDIX

Appendix A

Appendix 1. Current age of participants in years (n=109).

Age	Percentage	Responses (n)
<30 years old	49.54%	54
30-39 years old	31.19%	34
40-49 years old	12.84%	14
50-59 years old	5.50%	6
>60 years old	0.92%	1
	Answered	109

Appendix 2. Percentage of prenatal practice focus from participants (n=109).

Percent of Practice	Percentage	Responses (n)
0%	0.92%	1
Less than 25%	10.09%	11
Around 50%	12.84%	14
More than 50%	76.15%	83
	Answered	109

Appendix 3. Participants' experience in years providing prenatal genetic counseling services (n=109).

Time (years)	Percentage	Responses (n)
Less than 1 year	17.43%	19
1-5 years	50.46%	55
6-10 years	13.76%	15
11-15 years	3.67%	4
More than 15 years	14.68%	16
	Answered	109

Appendix 4. Participant employer settings (n=109).

Employer	Percentage	Responses (n)
Public hospital or clinic	65.14%	71
Private practice	21.10%	23
Genetics laboratory	8.26%	9
A company that contracts genetic counseling services	1.83%	2
Other (please specify)	3.67%	4
	Answered	109

Appendix 5. How often are prenatal genetic counselor's providing pretest counseling for NIPT (n=109).

Frequency of pretest counseling	Percentage	Responses (n)
None of the time	4.59%	5
Some of the time	69.72%	76
Most of the time	25.69%	28
All of the time	0.00%	0
	Answered	109