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# What Women Want: Lead Considerations for Current and Future Applications of Noninvasive Prenatal Testing in Prenatal Care

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## Abstract

**Background—**Noninvasive prenatal testing (NIPT) will change the delivery of prenatal care for all women, including those considered low-risk for fetal chromosomal abnormalities. This study investigated pregnant women's attitudes, informational needs, and decision-making preferences regarding current and future applications of NIPT.

**Methods—**A survey instrument was used to identify aspects of the decision-making process for NIPT among low-risk and high-risk populations.

**Results—**Both low-risk and high-risk women (n=334) expressed interest in incorporating NIPT as a screening test into their prenatal care. Information specific to NIPT's detection rate (86%), indications (77%), and performance in comparison with conventional screens and diagnostic tests (63%) were identified as lead factors when considering its use. The future availability of NIPT as a diagnostic test increased women's willingness to undergo testing for fetal aneuploidy, cancer susceptibility, childhood-onset and adult-onset diseases. Despite its noninvasive aspects, participants expressed the need for a formal informed consent process (71%) to take place prior to testing.

**Conclusions—**Our study demonstrates that NIPT will introduce new challenges for pregnant women and their healthcare providers who will be charged with supporting informed decision-making about its use. It is critical that obstetric professionals are prepared to facilitate a patient-centered decision-making process as its clinical application rapidly changes.

### Keywords

prenatal genetic testing; maternal decision-making; informed consent

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## INTRODUCTION

Noninvasive prenatal testing (NIPT) is a new form for prenatal screening changing the delivery of prenatal care for all women, including those traditionally considered as low-risk for fetal aneuploidy. NIPT measures cell-free fetal DNA in a pregnant woman's blood to provide a risk assessment for fetal chromosomal aneuploidy, specifically Trisomy 21, 13, and 18 (1, 2). The advantage of NIPT is that it can performed as early as 9-10 weeks of gestation and provides a higher detection rate coupled with a lower false positive rate than conventional first or second trimester screens (3-5). However, it has a 1% false positive rate and follow-up invasive diagnostic testing is necessary to confirm the presence or absence of aneuploidy. Furthermore, the screen does not provide information about the chance other fetal abnormalities that may be provided by using convention screens (e.g. neural tube defects) (2,6,7). At the present, NIPT is primarily recommended for women of advanced maternal age (2, 4, 8, 9), as the test's sensitivity and specificity in low-risk populations has not been well established. Over time, however, with advances in the science of cell-free fetal DNA (10-14), it is expected that NIPT will evolve into a diagnostic tool that can generate detailed genetic information about the fetus while bypassing the risks of invasive testing and the uncertainties of screening (15-18).

While NIPT offers clear advantages over conventional screens, there are important considerations about its use. Because fetal genetic information gained from NIPT can initiate a chain of decisions with an indelible impact on the current pregnancy and future reproductive decision-making (19-21), it is critical that a pregnant woman receives the necessary resources from her healthcare provider to make meaningful and informed choices about incorporating this new test into prenatal care. These resources must provide the educational and decision-making support needed to amass knowledge not only about indications, advantages, and disadvantages of NIPT but also other first and second trimester testing options to navigate all of her choices in an informed manner (3, 8-11). Such resources should also provide the opportunity to explore personal values and beliefs about genetic testing, disability, parenthood, and abortion and in addition to the tools to individualize risk information gained from NIPT (22-24).

As NIPT is an emerging technology, it is unclear how women conceptualize its risks and benefits as part of their prenatal care. Furthermore, it is unknown how these factors will change as NIPT becomes widely available as a screen among low-risk populations and to all women as a form of diagnostic testing. This information is central to patient-centered maternal care as prenatal genetics plays an increasing role in its delivery. The objective of this study was to identify pregnant women's attitudes, informational needs, and decision-making preferences regarding the evolving applications of NIPT, data key to structuring clinical practices around its integration into prenatal care.

## METHODS

A cross sectional study was conducted to identify pregnant women's attitudes, educational needs, and decision-making preferences regarding NIPT. Participants were recruited from the patient population of women presenting for outpatient prenatal care at the Cleveland

Clinic's Women's Health Centers. Eligible participants included women **in any trimester of pregnancy**, 18-45 years of age, that could read and speak English, and provide consent for participation. Patients who met these criteria were provided with a study packet as they checked in for their prenatal care visit. Study packets consisted of a cover sheet explaining the purpose of the study, informed consent procedures, and a self-administered anonymous questionnaire to be completed prior to their prenatal visit. All procedures and materials were approved by the Cleveland Clinic Institutional Review Board prior to study initiation.

The questionnaire was developed in conjunction with experts in clinical genetics, maternal-fetal medicine, ethics, and medical decision-making. It consisted of a series of Likert-scale responses, and multiple choice questions. Questions were preceded by an explanation of prenatal testing options, including a description of chorionic villus sampling (CVS), amniocentesis, and NIPT. Items were based on the authors' prior qualitative and quantitative research regarding women's use of novel prenatal tests. The instrument was piloted in a prenatal clinic setting and modified based on the responses of participants prior to field use.

The survey was divided into three sections; each section focused on a component of the decision-making process pertinent to current (e.g. screening) and future (e.g. diagnostic tool for single mutations and genomic analysis) applications of NIPT. Section 1 consisted of a series of multiple-choice items to examine the participants' decision-making needs and preferences for NIPT. Section 2 examined issues specific to genetic counseling and informed consent practices for NIPT. Section 3 examined attitudes about undergoing prenatal genetic testing for a variety of different genetic conditions.

Survey data was analyzed using SPSS version 18.0. Categorical variables were summarized by frequency and percentages for the total sample, and by age-related risk status (high-risk, i.e. 35 years or older at the time of delivery/advanced maternal age vs. low-risk, < 35 years of age), educational status, race/ethnicity, and whether the participant had any form of genetic screening or diagnostic testing during the current or a prior pregnancy. Testing of differences between groups was performed using Pearson's chi-square test. Attitude responses assessed on the Likert scales were treated as binary variables with "Not at all likely" and "Unlikely" denoting an unfavorable response and "Likely" and "Very likely" denoting a favorable response. Significance for all analyses was determined by a p-value less than or equal to 0.05.

## RESULTS

A total of 334 questionnaires were completed. The mean age of participants was 30.8 years, at the upper end of the low-risk category (Table 1). The majority were in the second or third trimester of pregnancy. Most participants reported having prior pregnancies (65.4%). Racial distribution of the participants was 77.7% white and 22.3% non-white (including 16.4% African American). Just over half (57.3%) of the participant population had completed college or graduate school, while 27.7% had completed some college work. Just under one-third of the participants (30.2%) had received genetic screening or diagnostic testing in the current or prior pregnancy.

## Current Application of NIPT as a Screening Test

Most participants (77.8%) prioritized the need to understand the indications of NIPT; specifically, which genetic conditions it could and could not help identify. Participants also expressed the need to know about the health and expected quality of life of a child born with Trisomy 21, 13, or 18 before initiating NIPT (61.1%). Other lead decision-making factors included the detection rate of NIPT (85.6%) and how it compares with conventional screens and diagnostic tests (62.6%). The likelihood and meaning of a false positive result was also an important factor (75.4%). Group differences were noted as high-risk women prioritized information about the chance of a false positive (88.3% vs. 75.0%, p=0.04). The chance of a false positive was also a concern of women who had prior screening or testing (82.5% vs. 68.8%. p=0.018) as well as how NIPT compares to other prenatal genetic tests (73.2% vs. 60.9%, p=0.05).

Nearly half of the respondents (42.6%) expressed an interest in having access to NIPT as early as possible in the pregnancy, a factor more common in high-risk women (57.9% vs. 40.6%; p=0.05) and those who had prior screening or testing (59.8% vs. 36.3%, p=0.002). Half of the respondents (50.6%) expressed an interest in obtaining information about the indications, benefits, limitations, and alternatives of NIPT at the time of their first prenatal visit. In addition, 32.0% expressed an interest in obtaining information about NIPT during preconception counseling or other clinical opportunities in advance of the first prenatal care visit.

The novelty of NIPT did not seem to make a significant difference in participants' perception about the time allotted to their decision making process. When considering NIPT, 81.0% reported they would be able **to decide whether to undergo NIPT** in the same time frame as conventional screens. No differences were noted by prior screening or genetic testing, but racial and educational differences were noted. White women with a college education or more were more likely to require <24 hours to decide whether to have NIPT compared to white women with less education (65% vs. 35%; $p < 0.05$) who required ≥24 hours. In contrast, when considering how long they would need to make a well-informed decision about amniocentesis or CVS, 60.2% of participants reported that they would need ≥ 24 hours because of the invasiveness and inherent risks of these procedures. No group differences were noted.

## Evolving Applications of NIPT

In anticipation of forthcoming applications of this technology, participants were queried about preferences regarding fetal genetic testing with NIPT compared to CVS and amniocentesis. Statistically significant differences ($p < .0001$) were observed in the participants' willingness to undergo testing for a number of different genetic conditions if information about the fetus could be obtained using NIPT instead of invasive diagnostic procedures (Table 2). These differences included testing to identify fetal aneuploidy (64.3% versus 35.5%, $p<0.001$), mutations associated with childhood-onset diseases (71.6% versus 51.1%, $p<0.001$), and serious, adult-onset diseases (56.1% versus 37.6%, $p<0.001$). Non-white participants were more likely to report that they would undergo fetal genetic testing if NIPT could be used instead of invasive procedures to assess for Down syndrome (OR 2.5,

p=0.008), childhood-onset disease (OR 2.6, p=0.009), adult-onset disease (OR 11.1, p<0.001), cancer susceptibility (OR 10.5, p <0.001), and determining fetal sex (OR 4.9, p=0.003). Women who had prior screening or testing were also more likely to utilize NIPT over invasive testing for Trisomy 21 (OR 2.6, p<0.001), childhood-onset disease (OR 2.3, p=0.001), but less likely to utilize NIPT to determine fetal sex (OR 0.4, p=0.012). (Table 3)

### Structuring Informed Consent for NIPT

Participants were surveyed about needs and preferences regarding informed consent practices for NIPT. The majority (71.0%) believed that a formal informed consent process should take place prior to undergoing NIPT. Participants were queried about core issues related to informed consent, specifically the perceived ability to make voluntary choices about the use of NIPT. One-quarter (25.5%) reported that the noninvasive aspects of this new tool may lead to increased pressure to undergo prenatal genetic testing while 27.3% were unsure or could not exclude this possibility. No group differences by age or prior screening status were found.

## DISCUSSION

In this study, we set out to identify pregnant women's educational needs and decision-making preferences for NIPT. These findings represent important components of a complex and nuanced decision-making process about using NIPT to access fetal genetic information (25). On the one hand, the integration of NIPT returns attention to longstanding questions that are associated with conventional screens and diagnostic tests. Specifically, it highlights the ongoing need to overcome existing barriers to women's informed decision-making regarding prenatal genetic testing (20, 21, 26-29). On the other, NIPT poses new challenges to healthcare providers and pregnant women. In part, this may include a growing interest in prenatal genetic testing among women who might not otherwise have considered testing using invasive methods. An additional challenge will be ensuring that all women have the resources and support they need to make an informed choice about utilization of this test given the ease with which more accurate fetal genetic information can now be accessed.

Our study demonstrates that using NIPT as a diagnostic test may have a salient effect on pregnant women's overall attitudes about prenatal genetic testing. This observation is especially important as NIPT evolves into a diagnostic modality in the near future (8, 15-18, 30). Participants, who currently would not otherwise incorporate prenatal genetic testing into their prenatal care, would be willing to adopt NIPT. Respondents overwhelmingly accepted the use of NIPT for a number of genetic conditions, including fetal genetic markers for childhood and adult-onset diseases, cancer susceptibility, as well as fetal sex. Baseline age-related risk status did not have an important impact on the participants' interest in using NIPT. However, individual reproductive history may be another important predictor of women's interest in NIPT, a factor that would benefit from future investigation. This observation warrants further attention and may suggest that a concurrent shift in women's attitudes about the use of prenatal genetic testing may take place in conjunction with the evolution of NIPT. It may also reflect an evolution in how women view risks and consequences associated with obtaining fetal genetic information, given that this information

may be obtained noninvasively. Studies have shown that the availability and routinization of conventional prenatal tests affect women's experiences of pregnancy, regardless if they decline or accept testing (28, 31). What is not known is how women's needs, experiences, and perceptions of risk may shift in relation to NIPT as it becomes more widely accepted by healthcare providers and pregnant women. As a result, the medical community must prepare for expanded clinical applications of NIPT, not only in terms of its unique counseling and decision-making complexity, but also with regards to the potential for a larger and more diverse patient population seeking it as part of routine prenatal care.

The diagnostic capability of NIPT is not the only challenge to be addressed. As a screening test, NIPT presents specific and highly-nuanced decision-making considerations to potential users. While more accurate than **its** conventional screening counterparts, NIPT does have important limitations. Our study demonstrates that women prioritize information about the indications of NIPT, specifically which fetal genetic conditions the screen does and does not identify. This finding is critical as, while NIPT offers more accurate information about a specific set of aneuploidy conditions, it does not currently provide information about other genetic abnormalities which other screens and tests can help identify (2,6,7). This will be an important tradeoff that women must consider when weighing the advantages and disadvantages of NIPT over other testing options.

Furthermore, while NIPT has a lower false positive rate compared to the first trimester screen or quadruple screen, the possibility and implications of this scenario did not go unrecognized. In particular, respondents were interested in learning about the detection and false positive rate of NIPT, the diagnostic testing options available to them in the case of an elevated risk result, and the risks of fetal injury or miscarriage with follow-up invasive diagnostic tests. High-risk participants were more attuned to the concepts of test accuracy and screen positive rates compared to low-risk individuals. As the indications for NIPT are expanded to low-risk patients, it will be critical that all women are adequately educated about these core concepts of prenatal screening with which they may not be as familiar. Of note, these are the same concepts pregnant women have struggled with since the inception of prenatal screening. Studies show that, from the time of the initial integration of the triple and quadruple screens to the present, women have continued to encounter barriers related to health literacy, understanding concepts of risk, and obtaining patient-centered information about their prenatal testing options (22-24). Evidence of these barriers in the context of the first trimester screen, the most recent screen to be robustly integrated into routine prenatal care, highlights the challenges that will come with an expanding array of testing options (27, 32-36).

A significant finding of this study pertained to informed consent. Study findings demonstrate that women believe that informed consent should take place prior to undergoing NIPT. The significance of informed consent in noninvasive testing has been discussed by other authors (37, 38). Yet, obstetric healthcare providers demonstrate diverging views about the role of informed consent for NIPT and are less likely to believe informed consent should be obtained prior to its use (39). Our study findings, placed in the context of prior research, call for healthcare providers to understand how women conceptualize the utility, value and limitations of NIPT so that counseling and informed consent processes can be

structured around the needs and preferences of test users. In addition, practice patterns regarding informed consent for NIPT should also be revisited as, at present, a formal informed consent process is primarily reserved for test involving invasive procedures. Thus, the need for effective counseling and decision-making tools will increase with the further clinical integraton of NIPT, especially in the context of medical practice which currently faces limitations of time and resources in addition to rapid advances in genetic science.

Despite the importance of a formal informed consent process, participants did not anticipate needing more time to **decide whether or not to undergo** NIPT compared to conventional tests. They did, however, express a concern for the potential to feel increased pressure to undergo testing with the availability of NIPT. These findings suggest that pregnant women view the informed consent process as a way to make a deliberate decision about NIPT. Establishing effective informed consent practices now in the early stages of this test's introduction will set an important precedent as advances in cell-free fetal DNA technology foster the diagnostic capabilities of this test.

This study has provided important insight about the introduction of NIPT into prenatal care. However, we acknowledge that the study has some limitations that should be considered when generalizing the results to pregnant woman as a whole. As NIPT is in its early stages of clinical integration into prenatal care, the responses of participants may change once the screen becomes more widely available. Further studies will be needed to examine the findings among larger and more diverse populations and as the non-invasive platform for prenatal testing evolves. Although the findings may be limited to our sample population, the data provides important insight into the educational and decision-making components necessary for informed uptake of this new technology.

## CONCLUSIONS

In conclusion, this study demonstrates that, while the introduction of NIPT will introduce some new challenges for pregnant women and their healthcare providers, it will also renew existing challenges in the provision of genetic testing. As an anticipated diagnostic tool, NIPT may alter the way women view the landscape of prenatal genetic testing and its role in their prenatal care. These findings call for effective mechanisms to ensure pregnant women are prepared to make informed decisions about NIPT as part of a growing and complex decision-tree of prenatal testing options, including overcoming obstacles to informed decision-making that already exist with the utilization of conventional prenatal tests.

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## Table 1

Participant Characteristics

| Factor | Total N (%) | < 35 years n (%) | >35 years n (%) | p value |
|---|---|---|---|---|
| Age | Mean: 30.8 yr Range: 19-44 | 256 (81.0) | 60 (18.9.) | <.0001 |
| Gravidity | | | | |
| G0 | 111 (34.6) | 97 (38.3) | 10 (16.7) | 0.002 |
| G + | 210 (65.4) | 156 (61.7) | 50 (83.3) | |
| Trimester | | | | |
| 1st | 66 (20.6) | 51 (20.2) | 15 (25.0) | 0.70 |
| 2nd | 127 (39.7) | 101 (39.9) | 22 (36.7) | |
| 3rd | 127 (39.7) | 101 (39.9) | 23 (38.3) | |
| Race/Ethnicity | | | | |
| Asian | 7 (2.2) | 4 (1.6) | 3 (5.1) | 0.52 |
| African American | 53 (16.4) | 43 (16.8) | 8 (13.6) | |
| Hispanic | 1 (0.3) | 1 (0.4) | 0 (0) | |
| White | 251 (77.7) | 199 (77.7) | 46 (78.0) | |
| Other | 11 (3.4) | 9 (3.5) | 2 (3.4) | |
| Education | | | | |
| Some HS/HS grad or GED | 48 (15.0) | 40 (15.8) | 6 (10.0) | 0.069 |
| Some College | 89 (27.7) | 75 (29.6) | 9 (15.0) | |
| College graduate | 94 (29.3) | 71 (28.1) | 23 (38.3) | |
| Graduate or professional degree | 90 (28.0) | 67 (26.5) | 22 (36.7) | |
| Genetic screening or testing during current or prior pregnancy | 97 (30.2) | 72 (28.5) | 22 (36.7) | 0.43 |
| Genetic testing when not pregnant | 21 (6.6) | 16 (6.3) | 3 (5.0) | 0.87 |
| Genetic counseling at any time | 31(9.6) | 24 (9.4) | 6 (10.0) | 0.53 |

Note: Not all participants supplied demographic information

## Table 2

Comparison of Acceptance of Testing Options in the Total Sample

| **Non-invasive Prenatal Genetic Testing (n,%)** | | **CVS/Amniocentesis (n,%)** | | |
|---|---|---|---|---|
| **Very likely/Likely** | **Not Likely/Never** | **Very likely/Likely** | **Not Likely/Never** | **p value** |
| *How likely are you to have NIPT vs. CVS or amniocentesis to see if the developing baby has Down syndrome?* | | | | |
| 211 (64.3) | 95 (29.0) | 118 (35.5) | 189 (56.9) | <.0001 |
| *How likely are you to have NIPT vs. CVS or amniocentesis to see if the developing baby will be born with a serious, life threatening illness?* | | | | |
| 235 (71.6) | 75 (22.9) | 170 (51.1) | 140 (42.0) | <.0001 |
| *How likely are you to have NIPT vs. CVS or amniocentesis to see if the developing baby will develop a serious, life threatening disease an adult?* | | | | |
| 183 (56.1) | 112 (34.4) | 125 (37.6) | 178 (53.6) | <.0001 |
| *How likely are you to have NIPT vs. CVS or amniocentesis to see if the developing baby will have an increased risk of developing cancer in the future?* | | | | |
| 165 (50.6) | 126 (38.5) | 90 (27.0) | 210 (63.1) | <.0001 |
| *How likely are you to have NIPT vs. CVS or amniocentesis to see if the developing baby is male or female?* | | | | |
| 161 (49.4) | 125 (38.3) | 64 (19.3) | 244 (73.7) | <.0001 |

**Table 3**

Acceptance of Invasive Prenatal Diagnosis and NIPT for Specific Conditions: Comparisons by Race, Education and Prior Screening or Genetic Testing (%)

| Factor | Down Syndrome | | Childhood Onset Disease | | Adult Onset Disease | | Cancer Susceptibility | | Fetal Sex | |
|---|---|---|---|---|---|---|---|---|---|---|
| | CVS/Amnio | NIPT | CVS/Amnio | NIPT | CVS/Amnio | NIPT | CVS/Amnio | NIPT | CVS/Amnio | NIPT |
| Total Sample‡ | 35.3 | 63.2 | 50.9 | 70.4 | 37.4 | 54.8 | 26.9 | 49.4 | 19.2 | 48.2 |
| White | 30.3 | 61.0 | 45.8 | 67.7 | 31.5 | 49.4 | 19.1 | 43.4 | 14.3 | 45.4 |
| Non-white | 50.6* | 69.9* | 66.3* | 78.3# | 55.4† | 71.1† | 50.6† | 67.5† | 33.7† | 56.6# |
| ≤High school | 38.7 | 62.0 | 51.8 | 67.2 | 41.6 | 56.9 | 35.0 | 51.1 | 29.2 | 53.3 |
| College or more | 31.5 | 65.8 | 48.9 | 73.9 | 33.2 | 54.9 | 20.1* | 50.0 | 10.9† | 46.2 |
| **No prior screen/testing** | **28.3** | **57.3** | **46.0** | **67.2** | **35.7** | **54.5** | **26.6** | **48.8** | **23.3** | **49.4** |
| **Prior screen/testing** | **50.6†** | **87.1†** | **66.7*** | **88.3†** | **46.0** | **71.9*** | **29.8** | **68.9*** | **10.2#** | **66.3#** |

Note:

Subgroup comparisons: p values from chi square - difference between demographic categories (white vs. non white; high school education vs. college education or more, **prior screening or genetic testing vs. not)**

‡Total sample: percentages of total participants who provided demographic information and as such, differ slightly from %'s presented in Table 2; p = <.0001 for all comparisons of acceptance of invasive testing vs. NIPT in the total sample population;

†p <0.001

*p<0.01

#p<0.05