The Changing Landscape of Prenatal Testing: Certified Nurse-Midwives’ Integration of NIPT into Practice

Master’s Thesis

Presented to

The Faculty of the Graduate School of Arts and Sciences
Brandeis University

Graduate Program in Genetic Counseling
Judith Tsipis, PhD, Advisor

In Partial Fulfillment
of the Requirements for the Degree

Master of Science
in
Genetic Counseling

by
Lisa S. Weingarten

August 2016
Copyright by

Lisa S. Weingarten

© 2016
ACKNOWLEDGEMENTS

I would like to thank my advisor, Judith Tsipis, for all of her help, encouragement and support during this research project, from the development of the idea through to the completion of the written thesis. I would also like to thank my committee members, Jennifer Hume Helgeson and Gwen Ladentresse, for their enthusiasm for this research, helpful comments, and feedback. Thank you to Margarita Corral for her assistance with numerous aspects of the study, from survey development and refinement to data analysis and navigating SPSS.

To the members of the Brandeis genetic counseling program - Judith, Gretchen, Guy, Missy, Cassie, Janet, David, and Joe - and the rest of the faculty, lecturers, and guest speakers, thank you for your insight and assistance in helping me grow professionally and personally throughout these two years.

Thank you to my family, friends, and classmates who were with me during the program to provide love and support through difficult times and successes. Finally, thank you to my boyfriend, Vlad, for your presence, encouragement, space at your bay, and opportunities to procrastinate together.
ABSTRACT

The Changing Landscape of Prenatal Testing: Certified Nurse-Midwives’ Integration of NIPT into Practice

A thesis presented to the Graduate Program in Genetic Counseling

Graduate School of Arts and Sciences
Brandeis University
Waltham, Massachusetts

By
Lisa S. Weingarten

Non-invasive prenatal testing (NIPT) has been clinically available since 2011, however, there has been little research investigating how Certified Nurse-Midwives (CNMs) include this new testing option in their practice. The purpose of this study was to learn whether CNMs offer NIPT, to whom they offer it, details about their pre- and post-test counseling, and challenges they have encountered since introducing NIPT to their patients. We distributed an anonymous, online mixed-methods survey to 9,500 members of the American College of Nurse-Midwives’ e-mail list. Of 502 respondents meeting inclusion criteria, we found that 90% of CNMs are offering NIPT to at least some of their patients, and of these, 38% offer NIPT to all of their patients and 58% offer testing to low risk women at least some of the time. When offering
testing, 80% of respondents provide pre-test counseling often or always. Respondents ranked the following topics as most important to include in pre-test counseling: NIPT is only a screening test; it cannot detect all chromosomal conditions; and its detection rate is higher than that of maternal serum screening. More than half of CNMs who have received a positive NIPT result provided post-test counseling to their patients themselves, while the others primarily referred to Maternal Fetal Medicine specialists or Genetic Counselors. Major challenges identified by respondents included: the absence of consistent professional guidelines and clear insurance reimbursement policies for NIPT; the difficulty counseling patients regarding positive or indeterminate results, including incidental findings; and the struggle to stay up-to-date with advances in NIPT. Our results demonstrate that while almost all CNMs offer NIPT to patients, not all are confident in their knowledge of NIPT or how to discuss results and their implications with patients, signaling a need for further education and guidance. Genetic Counselors are well suited to help address some of these challenges.

Keywords: certified nurse-midwives, non-invasive prenatal testing, genetics, pre-test counseling, post-test counseling, education, genetic counseling.
# TABLE OF CONTENTS

Acknowledgements ........................................................................................................ iii

Abstract ........................................................................................................................... iv

Table of Contents ........................................................................................................... vi

List of Tables ................................................................................................................. viii

List of Figures ................................................................................................................ viii

Introduction ................................................................................................................... 1

Materials and Methods ............................................................................................... 6

Participants .................................................................................................................... 6

Instrumentation ............................................................................................................. 6

Procedures .................................................................................................................... 7

Data Analysis ................................................................................................................ 7

Results .......................................................................................................................... 8

Demographics .............................................................................................................. 8

NIPT Offering Practices ............................................................................................... 8

Pre-test counseling ....................................................................................................... 11

Post-test counseling ..................................................................................................... 14

Provider education and referrals ................................................................................. 15

Confidence in genetic counseling topics ..................................................................... 19

Discussion .................................................................................................................... 21

NIPT offering practices ............................................................................................... 21

High-risk versus low-risk populations ....................................................................... 22

Pre and post-test counseling ....................................................................................... 24

The impact of insurance coverage ............................................................................. 26
LIST OF TABLES

Table I: Qualitative analysis of desired education .............................................. 18
Table II: Confidence in Genetic Counseling Topics .............................................. 20

LIST OF FIGURES

Figure 1: Prenatal genetic testing options offered by CNMs .................................... 9
Figure 2: NIPT offering practices .......................................................................... 10
Figure 3: Frequency of providing pre-test counseling for NIPT ................................. 12
Figure 4: Topics included in pre-test counseling for NIPT ..................................... 13
Figure 5: Resources provided during pre-test counseling ....................................... 14
Figure 6: Positive NIPT results and post-test counseling practices ......................... 15
Figure 7: Resources to learn about NIPT .............................................................. 16
INTRODUCTION

Certified Nurse-Midwives (CNMs) are licensed advanced practice nurses who provide health care to women throughout their reproductive life span, including preconception, pregnancy, childbirth, and the post-partum period, as well as regular gynecological care and physical exams. The American College of Nurse-Midwives (ACNM) reported that as of February 2015, there were 11,018 CNMs in the US, who attend to over 12% of all vaginal births and 8.2% of all births in the country (ACNM, 2015). The proportion of CNM-attended births more than doubled since 1992, and their role in providing prenatal care is continuing to increase (ACNM, 2012). According to the Midwives Alliance of North America (MANA), a core competence of all midwives is understanding and describing the screening methods and diagnostic tests often used during pregnancy, including indications, risks and benefits of each (MANA, 2014). As new prenatal genetic tests are developed, CNMs must stay abreast of these advancing technologies and provide their patients with up-to-date testing options and information about each of them. This is especially important in geographical areas where genetics specialists, such as genetic counselors, are less available to counsel patients about these testing options.

While CNMs feel that understanding and explaining genetic concepts is important for their practice, especially as the field of prenatal screening advances, research has shown that nurses and advanced nurse practitioners, including CNMs, have both poor knowledge of genetic concepts, and poor self-reported confidence in this knowledge (Skirton, O'Connor, & Humphreys, 2012). Crane et al. surveyed 612 CNMs in the US, having them rank the importance
of, and their confidence in, various genetic and psychosocial aspects of their practice (Crane, Quinn Griffin, Andrews, & Fitzpatrick, 2012). They found that most CNMs felt integrating genetic information into practice was very important or essential; however, they only had moderate confidence in their abilities to understand and explain this information, compared with high confidence in their abilities to provide psychosocial counseling and support to patients (Crane et al., 2012). Furthermore, 98% of these CNMs wanted additional information on these concepts, signifying a need for future education about both genetics and prenatal screening, and a potential role for genetic counselors in this education (Crane et al., 2012). Tenby et al. surveyed Swedish midwives regarding prenatal testing practices and found that 83–89% of participants reported insufficient or no education regarding different prenatal tests and 94% of respondents desired more information about Down syndrome and prenatal testing (Ternby, Ingvoldstad, Anneren, & Axelsson, 2015). Of a sample of healthcare providers including MDs, NPs, RNs, and CNMs in the US, Sayers et al. found that 85% thought genetic counseling is a necessary part of prenatal testing, yet previous studies suggest that comprehensive genetic counseling is not routinely performed by CNMs in a prenatal setting throughout multiple different countries (Crane et al., 2012; Sayres, Allyse, Norton, & Cho, 2011; Ternby et al., 2015; Tomatir, Ozsahin, Sorkun, Demirhan, & Akdag, 2006).

New advances in non-invasive screening for fetal chromosomal abnormalities are revolutionizing the field of prenatal screening. Lo et al. first reported that cell-free ‘fetal DNA’ can be found circulating in the maternal blood stream in 1997, suggesting potential implications or application for non-invasive prenatal screening for chromosome abnormalities by analyzing the mother’s blood (Lo et al., 1997). These fragments of cell-free DNA originate from the placenta, rather than the fetus itself, and massively parallel sequencing (MPS) of maternal
plasma can accurately detect several aneuploidy pregnancies (Trisomy 21, 18, and 13) as early as 10 weeks gestation, with a detection rate and false positive rate of 99.2% and 0.09%, respectively, for Trisomy 21, 96.3% and 0.13% for Trisomy 18, and 91.0% and 0.13% for Trisomy 13 (Chan et al., 2004; Chiu et al., 2008; Gil, Quezada, Revello, Akolekar, & Nicolaides, 2015; Palomaki et al., 2012; Tjoa, Cindrova-Davies, Spasic-Boskovic, Bianchi, & Burton, 2006). A positive result from NIPT also carries a higher positive predictive value than a positive result from traditional combined first trimester screening (nuchal translucency measurements and maternal serum analyte analysis) for many conditions, including T21, T18 and T13 (Palomaki et al., 2012). Advances in NIPT technology are increasing the number of conditions screened for and now include sex aneuploidies, such as Klinefelter and Turner syndrome, as well as a variety of microdeletions. The new MaterniTGenome test, offered by Sequenom, is able to screen for fetal copy number variants larger than 7Mb in size on a genome wide scale (Bianchi et al., 2012; Lefkowitz et al., 2016; Srinivasan, Bianchi, Huang, Sehnert, & Rava, 2013; Wapner et al., 2015).

As the menu of testing options for NIPT is expanding, there is concern regarding whether providers, including CNMs, understand and can explain the conditions screened for by this test. Research has shown that the information physicians and midwives have regarding conditions other than T21 is poor (Maradiegue, Edwards, Seibert, Macri, & Sitzer, 2005; Metcalfe, Haydon, Bennett, & Farndon, 2008; Swaney, Hardisty, Sayres, Wiegand, & Vora, 2015). Despite this lack of understanding, obstetricians in the U.S. are in favor of offering NIPT to women to screen not just for T21, but for many additional chromosome aneuploidies, and many are either currently offering or plan to offer this ‘expanded’ testing to women in the future (Benn et al., 2014; Sayres et al., 2011). No studies have yet examined the conditions for which CNMs are offering testing or their confidence in explaining or describing these conditions.
Studies are ongoing to determine the performance of NIPT in women at low risk for fetal aneuploidy, and there is no consensus between professional societies as to whether it is appropriate to routinely offer this testing to women at low risk. Currently, the International Society of Prenatal Diagnosis and the American College of Medical Genetics and advocate offering cell-free DNA screening to all pregnant women regardless of their risk status; the American Congress of Obstetricians and Gynecologists (ACOG) and the Society for Maternal and Fetal Medicine (SMFM) do not explicitly suggest against offering it to low-risk women, but emphasize that positive predictive value is much lower in low-risk women; and the National Society for Genetic Counselors (NSGC) does not recommend routine screening for aneuploidy using NIPT for low risk women at this time (ACOG, 2015, 2016; Benn et al., 2015; Devers et al., 2013; Gregg et al., 2016).

Many physicians, however, seem to be comfortable offering NIPT to both high- and low-risk women; studies by Benn et al. and Musci et al. showed that 76% and 79% of US obstetricians, respectively, are comfortable offering NIPT to low-risk women (Benn et al., 2014; Musci et al., 2013). Some genetic counselors, on the other hand, feel that NIPT should be reserved for high-risk populations and not used as a routine screening practice (Buchanan, Sachs, Toler, & Tsipis, 2014). There is currently little information in the literature about the thoughts and practices of CNMs regarding this issue.

Despite CNMs being involved in an increasing proportion of prenatal care in the United States, knowledge regarding how these providers use NIPT is lacking. The primary goals of this research study were to: i) determine if and to whom CNMs are offering NIPT, ii) explore CNMs’ experiences counseling patients about NIPT in both pre- and post-test counseling situations, iii) investigate their confidence in discussing various genetic counseling topics related to prenatal
testing and NIPT, and vi) determine some of the challenges experienced by CNMs as a result of integrating NIPT into their practice. This study provides information regarding how CNMs are incorporating NIPT into the prenatal services they offer to their patients and suggests ways genetic counselors and the labs offering NIPT can best support CNMs’ efforts to broaden their scope of prenatal screening.
MATERIALS AND METHODS

Participants

Participants for this research study were Certified Nurse-Midwives (CNMs) practicing in the United States who currently provide prenatal/antepartum care to women. These CNMs had email addresses on file with the American College of Nurse-Midwives (ACNM). We did not exclude based on age, gender, geographic location within the US, or other demographic characteristics. Although included in the ACNM email list, we excluded student members, Certified Midwives (CMs) and retired CNMs from participating in this study.

Instrumentation

We developed an anonymous survey consisting of multiple choice, Likert scale, and short-answer/free response format questions (see Appendix A). The survey included the following sections: inclusion/exclusion screening questions, assessment of whether CNMs offer NIPT, the patient populations to which CNMs offer testing, topics discussed with patients during pre- and post- test counseling, referral practices, and how CNMs are educated about NIPT. The survey concluded with two short answer questions to explore in more depth the challenges CNMs face as they are offering NIPT in their practices and further education desired, followed by basic demographic questions including age, gender, years in practice, type of employer (ie hospital, community medical center), and patient demographics.
**Procedures**

To recruit a sample of eligible CNMs, we sent a recruitment email to approximately 9500 ACNM members with email addresses on file. This recruitment email described the survey and inclusion criteria, and contained a link to the anonymous online survey (see Appendix B). We created and hosted the survey using Qualtrics®, an online survey tool. Participation in this study was voluntary and entry into the survey presumed participant consent. Respondents had the option to withdraw at any time during the survey or skip any question. As an incentive to participate in the survey, we offered respondents the opportunity to enter a raffle for one of three $50 Amazon gift cards by providing their email address in an unlinked Qualtrics survey. The ACNM distributed the initial recruitment email on 3/31/16 followed by an email reminder on 4/12/16, and we closed the survey on 4/19/16. This study was approved by the Brandeis Institutional Review Board (IRB) (protocol #16073) and the ACNM approved the solicitation of CNM/CM members via their email list.

**Data Analysis**

We performed quantitative analysis with SPSS version 23. We calculated descriptive statistics to analyze the demographics of the sample. For other survey items, we first calculated descriptive statistics for each item and then used chi-square analysis, t-tests, correlations, and cross tabulations to identify significant or interesting associations between survey items. We set statistical significance at $p = 0.05$. We manually analyzed responses to open-ended questions in Microsoft Excel using an inductive approach to identify common themes related to CNMs’ experiences and feelings regarding the implementation of NIPT into their practice, challenges faced, and the type of further education they desire.
RESULTS

We received 583 total responses and removed 81 responses from our analysis due to failure to complete the majority of the survey ($n=47$) or failure to meet inclusion criteria ($n=34$). We analyzed data from 502 respondents, which equates to approximately a 5% response rate based on the estimated number of ACNM members to which the survey was distributed (9500); however, not all ACNM members met our inclusion criteria so the response rate of those eligible to participate is greater than 5% but cannot be readily calculated.

Demographics

The majority of respondents were female (99.4%, $N=500$), with a mean age of 49.7 years ($SD = 11.7$, $N=494$) and an average of 16.5 years of experience working as a CNM ($SD = 10.2$, $N=483$). The most common primary employers were hospitals/medical centers (36%), followed by physician-owned practices (27%) and midwifery-owned practices (14%) ($N=501$). Respondents practiced throughout the United States, with approximately equal distribution between those practicing in the North East, Midwest, South and West (28%, 19%, 29%, and 24% respectively, $N=502$). Fifty-six percent ($N=501$) of respondents see over 25 patients per week and 64% ($N=501$) indicated that between 75-100% of their patients have medical insurance, most commonly private health insurance or Medicaid.

NIPT Offering Practices

Ninety-one percent ($N=501$) of respondents reported that they offer NIPT to their patients at least some of the time. Screening options are offered more frequently to patients than
diagnostic options (CVS and amniocentesis), with maternal serum screening (first screen, sequential, integrated, or quad screen) and second trimester level II ultrasound offered to at least some patients by 98% and 96% of midwives respectively (N=498) (Figure 1). Whether or not respondents reported offering NIPT to their patients, 85% (N = 502) have referred patients to a genetic counselor to discuss this testing option.

**Figure 1: Prenatal genetic testing options offered by CNMs**

![Genetic Screening/Diagnostic Tests](chart)

* First screen, sequential, integrated, or quad screen. CVS = Chorionic villi sampling

Of the CNMs offering NIPT to their patients, 38% offer this testing to all patients, while 52% offer it to only some of their patients (N=501) (Figure 2a). When midwives offer NIPT to only some of their patients, 95% (N=262) offer NIPT to all high-risk women and they either do not offer it to low-risk women (72%, N=262), or only offer it to low-risk women if these patients bring up this testing option themselves (13%) (Figure 2a). Overall, 58% (N= 450) of CNMs who offer NIPT have offered this test to low-risk women, while 42% have not (Figure 2b).
The most common reasons that CNMs do not offer NIPT to any of their patients were (N=52): i) the fact that NIPT is not offered at their institution by any provider (33%) ii. that testing is not always covered by insurance (27%), and iii) that other providers at their institution
are responsible for ordering this test (25%). Confidence in describing NIPT and interpreting results were each selected a reason for not offering NIPT by 8% of respondents. One third (N=51) of respondents who do not currently offer NIPT are planning to offer it to their patients in the future, and another 53% reported that they were unsure whether this test will become a part of their future offerings.

**Pre-test counseling**

Eighty percent (N=450) of respondents who offer NIPT reported that they often or always provide a pre-test counseling discussion to their patients (Figure 3). Furthermore, 44% (N=350) of CNMs who provide pre-test counseling often or always still refer to a genetic counselor to discuss NIPT prior to testing. There was no statistically significant relationship between how often CNMs provide pre-test counseling and their patient load, region, or years of practice as a CNM. However, CNMs in a midwifery-owned practice were significantly more likely to report providing pre-test counseling often or always compared with those who work in other settings (92%, N=62 versus 78%, N = 387, p < 0.01).
When asked to indicate the five most important topics to include in a pre-test counseling discussion (N=434) the following three topics were each chosen by over 50% of respondents: “NIPT is only a screen; it is not diagnostic (false positive and false negative results can occur)”, “NIPT cannot screen for all chromosome abnormalities or genetic conditions”, and “NIPT has better detection rates and lower false positive rates than maternal serum screening” (Figure 4). Only 24% of respondents included a description of Down syndrome in their selections, while 48% chose a description of conditions OTHER than Down syndrome tested for by NIPT. The two topics chosen least frequently were discussing no-call or indeterminate results (14%) and incidental findings (7%).

Figure 3: Frequency of providing pre-test counseling for NIPT
Figure 4: Topics included in pre-test counseling for NIPT

We asked respondents to indicate which types of resources they provide to their patients during pre-test counseling discussions; the majority selected printed materials/handouts, with those produced by an NIPT testing company being used slightly more frequently than those not produced by a testing company (47% versus 43%, N= 432) (Figure 5).
Post-test counseling

At least 61% \((N = 449)\) of CNMs offering NIPT to patients have had one or more patients screen positive on this test (Figure 6a). Two thirds \((N = 272)\) of respondents who have experienced a positive NIPT result provided post-test counseling themselves after receiving a positive result, while the remainder primarily referred patients to Maternal Fetal Medicine specialists \((n = 59)\) and/or Genetic Counselors \((n = 70)\) (Figure 6b). Down syndrome was the most commonly experienced positive NIPT result \((80\%, N = 269)\), followed by Trisomy 18 \((42\%, N = 269)\) (Figure 6c). There was no statistically significant relationship between whether or not CNMs provided post-test counseling to patients receiving a positive NIPT result and patient load, region, type of employer, or years of experience as a CNM.
Provider education and referrals

CNMs are educated about NIPT through a variety of sources. When asked to select the three most helpful resources to learn about NIPT from the list provided, about 40% of respondents chose each of the following options: talks presented by a lab company, directed review of the literature, and resources provided by a professional organization (Figure 7).
Furthermore, 31% of CNMs thought consulting with a genetic counselor was most helpful (Figure 7).

**Figure 7: Resources to learn about NIPT**

<table>
<thead>
<tr>
<th>Which 3 resources have been most helpful to learn about NIPT?</th>
<th>Percent</th>
</tr>
</thead>
<tbody>
<tr>
<td>Educational talks presented by a lab company</td>
<td>40.8%</td>
</tr>
<tr>
<td>Self-directed review of literature</td>
<td>40.2%</td>
</tr>
<tr>
<td>Resources provided by a professional organization</td>
<td>39.4%</td>
</tr>
<tr>
<td>Consulting with non-GC peers or co-workers</td>
<td>32.4%</td>
</tr>
<tr>
<td>Consulting with a GC</td>
<td>30.8%</td>
</tr>
<tr>
<td>Literature/brochures produced by a lab company</td>
<td>27.8%</td>
</tr>
<tr>
<td>Educational talks NOT presented by a lab company</td>
<td>20.6%</td>
</tr>
<tr>
<td>Presentations or events at ACNM meeting</td>
<td>11.4%</td>
</tr>
<tr>
<td>Formal education/CNM degree program</td>
<td>9.0%</td>
</tr>
<tr>
<td>Literature/brochures NOT produced by lab company</td>
<td>8.0%</td>
</tr>
<tr>
<td>Other source</td>
<td>4.6%</td>
</tr>
<tr>
<td>I have not received useful information about NIPT</td>
<td>3.2%</td>
</tr>
</tbody>
</table>

*N=500*

We asked respondents to list the one topic they would be most interested to learn about in a hypothetical lecture on NIPT. Using a manual inductive approach, we grouped their short answer responses into four themes (Table II):

1. **Communicating results**: how to discuss positive as well as ‘non-typical’ results, and details about no-call/indeterminate results and incidental findings;
2. **Advancing science and technology**: learning more about how NIPT works, where the technology may be going, potential future uses for non-invasive testing;
3. **Practical/logistical considerations**: determining if a patient’s insurance will cover testing or what to do if patients do not have health insurance, as well as learning about how different testing laboratories’ offerings compare in terms of tests available, accuracy, and cost; and

4. **Predictive value and accuracy**: the sensitivity and specificity of NIPT for various conditions and age groups, false positives and negatives, and the utility of this testing in low-risk populations.
Table I: Qualitative analysis of desired education

If you were to attend one lecture about NIPT, what topic would you be interested to learn more about?

<table>
<thead>
<tr>
<th>Theme</th>
<th>Subthemes</th>
<th>Examples</th>
</tr>
</thead>
<tbody>
<tr>
<td>Communicating Results</td>
<td>Counseling for different results</td>
<td>“Counseling the non-negative result: positives, indeterminates [<em>sic</em>] and maternal conditions.”&lt;br&gt;“How to handle different possible results from the testing and where to go for additional resources.”</td>
</tr>
<tr>
<td></td>
<td>Incidental and indeterminate findings</td>
<td>“Other incidental findings, especially maternal conditions, that can be found with NIPT.”&lt;br&gt;“Microdeletions, monosomy, triploidy, etc”&lt;br&gt;“No call/indeterminant [<em>sic</em>] results and incidental findings stats”</td>
</tr>
<tr>
<td>Advancing science and technology</td>
<td>Future directions of non-invasive testing</td>
<td>“why is it not more broad spectrum and can it be in the future?”&lt;br&gt;“Future used of NIPT. I.e. RH typing etc.”</td>
</tr>
<tr>
<td></td>
<td>How NIPT works</td>
<td>“I would love to learn more about the technology of the testing”&lt;br&gt;“How the test works, the info it provides…”</td>
</tr>
<tr>
<td>Practical /logistical considerations</td>
<td>Cost and insurance coverage</td>
<td>“access for uninsured, undocumented patients”&lt;br&gt;“how much does it cost, and does insurance pay for it. because few if any of my patients would pay for it themselves.”</td>
</tr>
<tr>
<td></td>
<td>Comparing different lab’s tests</td>
<td>“Comparisons between the different available tests, and cost effectiveness data for each”&lt;br&gt;“Head to head comparisons with competing lab tests”</td>
</tr>
<tr>
<td>Predictive Value</td>
<td>Accuracy and reliability of NIPT</td>
<td>“Accuracy across different age group / risk categories”&lt;br&gt;“comparing its sensitivity and specificity to sequential screen”&lt;br&gt;“efficacy rates of detecting anomalies and false positive rates”</td>
</tr>
<tr>
<td></td>
<td>Utility and validity in low risk women</td>
<td>“It's validity in low-risk patients. We've been counseled against using it in low-risk women by our MFM group but it seems most offices ignore this and order it for all women.”&lt;br&gt;“what to say to women not at high risk (under 34, no hx) - who are interested in this form of testing”</td>
</tr>
</tbody>
</table>
Confidence in genetic counseling topics

We asked respondents to rank their confidence in various genetic counseling topics on a scale from 1 to 5, with 1 being not confident at all and 5 being completely confident (Table III). On average, respondents were most confident in their ability to discuss diagnostic testing options to confirm screening results (CVS/amniocentesis), discuss the accuracy and reliability of NIPT, and facilitate decision making about genetic testing options. The lowest self-reported confidence scores were in the two condition-specific topics: discussing care and management for a child with Down syndrome/trisomy 21 and for a condition other than Down syndrome (ie T13, T18, sex chromosome aneuploidies, etc). Compared with those who do not offer NIPT, CNMs who offer NIPT to at least some patients reported higher levels of confidence in their ability to discuss diagnostic testing ($M=3.26$ versus 2.80, $p<0.01$) and termination after a positive genetic test ($M=3.79$ versus 3.31, $p<0.05$). Furthermore, those who provide pre-test counseling and those who provide post-test counseling were significantly more confident in every topic compared with those who do not provide counseling ($p<0.05$).
**Table II: Confidence in Genetic Counseling Topics**

<table>
<thead>
<tr>
<th>Discussion topic</th>
<th>N</th>
<th>M (SD)</th>
<th>% mostly or completely confident</th>
</tr>
</thead>
<tbody>
<tr>
<td>Diagnostic testing options to confirm screening results (cvs or amniocentesis)</td>
<td>497</td>
<td>3.74 (1.04)</td>
<td>64.0%</td>
</tr>
<tr>
<td>The accuracy and reliability of NIPT**</td>
<td>448</td>
<td>3.66 (0.90)</td>
<td>64.1%</td>
</tr>
<tr>
<td>Facilitating decision making regarding genetic testing options</td>
<td>499</td>
<td>3.57 (1.00)</td>
<td>58.7%</td>
</tr>
<tr>
<td>Counseling regarding the option to end a pregnancy after a positive (abnormal)</td>
<td>498</td>
<td>3.22 (1.31)</td>
<td>46.8%</td>
</tr>
<tr>
<td>genetic testing result</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Implications of a no-call/indeterminate result on NIPT**</td>
<td>448</td>
<td>2.79 (1.20)</td>
<td>29.7%</td>
</tr>
<tr>
<td>Care and management for a child with Down syndrome/trisomy</td>
<td>500</td>
<td>2.65 (1.22)</td>
<td>27.6%</td>
</tr>
<tr>
<td>Potential for incidental findings from NIPT**</td>
<td>448</td>
<td>2.65 (1.14)</td>
<td>23.9%</td>
</tr>
<tr>
<td>Care and management for a child with a chromosome condition <em>OTHER</em> than Down</td>
<td>500</td>
<td>2.10 (1.08)</td>
<td>12.2%</td>
</tr>
<tr>
<td>syndrome</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

**asked only to respondents who offer NIPT to at least some patients.**
Respondents were asked to rank confidence discussing each of the topics on a scale from 1 = not confident at all to 5 = completely confident. *M* = mean, *SD* = standard deviation. % mostly or completely confident = percent of respondents who indicated a confidence level of 4 or 5 for the specific discussion topic.
DISCUSSION

NIPT offering practices

Our study found that the vast majority (90%) of CNMs in the United States are offering NIPT to at least some patients, with 38% of respondents offering this testing to all of their patients and 52% offering it to some of their patients. Because this technology has been clinically available for only the past 5 years, it is not surprising that CNMs offer NIPT less frequently than maternal serum screening (MSS) and anatomical ultrasound. These results suggest that CNMs are not using NIPT to replace the more traditional screening tests, but rather are offering it in addition to these tests. All types of screening tests, including NIPT, were offered more often than diagnostic testing options (CVS and amniocentesis), which are invasive and carry a risk of miscarriage or other complications. Many respondents commented how NIPT is valuable to their patients as it is an accessible, accurate way to again information about a pregnancy without the risk of miscarriage associated with diagnostic tests:

“This has been a real change of practice for our small, rural clinic. Amnio was not available readily here and this has been a great way of women to feel more confident about their pregnancy.”

“[I] Feel that it is a great test which has revolutionized prenatal testing. Many high risk women who would not do testing in past due to fear of miscarriage now do testing.”

The midwives who do not currently offer NIPT cited practice structure as one of the main reasons for not offering the testing, rather than lack of understanding or confidence in explaining NIPT to patients or interpreting results. For example, other providers in their practice exclusively
offered this test or the practice as a whole did not offer this testing for a variety of reasons.

Consequently, when asked what changes would increase the probability they would offer NIPT to patients in the future, respondents listed changes in their practice structure and the accessibility of this testing more often than increasing their understanding of NIPT:

“If our lab and/our institution would offer it”
“Direct access through clinic rather than through genetics department”
“[If] other [sic] providers in my practice were doing it”

**High-risk versus low-risk populations**

When exploring CNMs’ NIPT offering practices, we found the proportion of CNMs who offer this screening test selectively to only some patients slightly exceeded those who offer it to all patients (60% vs 40% respectively). Comparing these groups, our results show that the majority of the midwives who offer NIPT to only some patients tend to offer it to all high-risk patients, and either not offer it to low-risk women, or only offer it to low-risk women if these patients bring up this option themselves. This data suggests that the CNMs who offer NIPT generally do so to all high-risk women, primarily differing in whether they offer it to low-risk women.

Professional society all recommend offering NIPT it to high-risk women as one option for prenatal screening, but guidelines regarding best practices for cell-free DNA screening in the general/low-risk population are more variable and are constantly evolving. In fact, in the time since we closed this survey, two major professional groups have revised their options regarding optimal testing strategies in the low-risk population. The American Congress of Obstetricians and Gynecologists (ACOG) and the Society for Maternal Fetal Medicine (SMFM) jointly published a new practice bulletin which emphasizes the lower positive predictive value of NIPT
in low-risk populations and states the decrease in accuracy in this population is “concerning”, but does not explicitly discourage offering NIPT to low-risk women (ACOG, 2016). This is in contrast to their position statement published less than one year earlier, stating “conventional screening methods remain the most appropriate choice for first-line screening for most women in the general obstetric population” (ACOG, 2015). The American College of Medical Genetics and Genomics (ACMG) also recently updated their view on the topic, publishing a new position statement stating NIPT is an appropriate screening option for all pregnant women, while previous recommendations from 2013 did not explicitly endorse the use of NIPT in low-risk women (Gregg et al., 2013; Gregg et al., 2016). Similarly, the International Society for Prenatal Diagnosis (ISPD) released a position statement in 2015 supporting NIPT as an appropriate first-line screen for all pregnant women (Benn et al., 2015). On the other hand, NSGC’s position, last updated in 2013, “does not currently support NIPT as a routine, first-tier aneuploidy screening test in low-risk populations” (Devers et al., 2013).

Like those of professional societies, respondents’ opinions and practices varied with regard to offering NIPT to women at low risk for fetal aneuploidy. Some also questioned the value of this testing compared with other screening options for this population, such as maternal serum screening and nuchal translucency measurements:

“I wish it were a direct option for everyone regardless of age or risk criteria. It seems silly to do first trimester screening and quad screening when we have the NIPT option.”

“There is also no real data showing an increased advantage for low risk pts and ACOG still recommends MSS and NT for low risk pts.”

“How valuable is it for low risk women and should we be offering it to them instead of the QUAD screen? Should this become the new screening standard for everyone?”

Information related to the use of NIPT in low-risk women was one of the main areas in which respondents desired further education. Furthermore, when we asked CNMs do not offer NIPT to
their patients to describe the changes that might increase the probability they would offer it in the future, many cited updated, clear guidelines supporting the use of NIPT for low-risk women:

“*When it is recommended for low risk women and/or becomes new standard of care.*”

“*Clear indications for its use in low-risk women*”

These results show that further study of the validity and predictive value of NIPT in low-risk women, as well as clear and cohesive recommendations for offering NIPT to this population, are essential to clarifying best practices and helping CNMs understand the most appropriate ways to use this technology with their patients.

**Pre and post-test counseling**

We found that 80% of CNMs who offer NIPT provide patients with a pre-test counseling discussion often or always. The content and depth of what is included in these pre-test counseling sessions is still unclear, but we found that CNMs thought that the most important topics to include in pre-test counseling were the limitations of this technology (it is only a screen and cannot detect all genetic conditions) and how it compares with other screening technologies, such as maternal serum screening. These coincide with topics genetic counselors have recommended providers discuss with all patients in pre-test counseling for NIPT (Sachs, Blanchard, Buchanan, & Bianchi, 2015). The genetic counselors’ recommendations also encourage providers to discuss the conditions being screened for, the need to confirm abnormal results with diagnostic testing, and the possibility of incidental findings – three topics which ranked much lower in perceived importance to the CNMs. However, the CNMs’ mean confidence levels for discussing incidental findings and the conditions tested for (Down syndrome and other conditions) were the lowest of all topics assessed. This gap in reported
confidence and understanding provides genetic counselors an opportunity to educate CNMs about these topics, ultimately increasing patient understanding and informed decision making.

Referrals to a genetic counselor directly can also be especially helpful to providing comprehensive pre-test counseling for the many available genetic testing options in the context of an initial prenatal visit. Overall, 85% of respondents have referred patients to a genetic counselor for counseling about NIPT (either pre- or post-test), and, even when respondents themselves reported providing pre-test counseling often or always, 44% still indicated that they had referred patients to discuss NIPT with a GC prior to testing. Some respondents commented on how they include referrals to genetic counselors in their practice:

“all pts getting NIPT would see the genetic counselor prior to doing test. I introduce the concept, the genetic counselor gives details / answers questions that come up.”

“The availability of NIPT Is a valuable tool in helping assessing potential [sic] genetic problems for our clients. However, anything outside of a normal result need to be referred to a genetic counselor for more in-depth counseling […]”

Furthermore, 30% of respondents ranked consulting with a Genetic Counselor within the top three most helpful resources to learn about NIPT. Together, these findings suggest that many CNMs recognize the unique and specialized role GCs can play in the education of patients and providers.

We found inconsistencies between perceived importance and confidence in discussing chromosomal aneuploidy conditions, such as Down syndrome, and potential patient needs. Sixty one percent of CNMs offering NIPT had experienced at least one positive NIPT result, of which 80% were a positive screen for Down syndrome; however, only 24% of CNMs ranked Down syndrome as one of the five most important topics to include in a pre-test counseling discussion. The high proportion of CNMs who have received a positive NIPT results emphasizes the importance of discussing these conditions in pre-test counseling to prepare patients for the
possibility of receiving a positive screen. Without knowledge of what a positive result could mean for the fetus, the woman, and her family, informed consent may not possible and the usefulness of the test is questionable. As one respondent wrote:

“I primarily believe the testing is useful only if the woman/family knows what they would plan to do with the information [...]”

Furthermore, almost two thirds of respondents provided post-test counseling to their positive patients themselves, yet confidence in discussing both Down syndrome and other conditions tested for on NIPT were among the lowest out of all topics assessed ($M = 2.65$ and $2.10$ respectively). Genetic counselors are an ideal resource to provide information regarding the conditions tested for and to help CNMs develop strategies to facilitate patient decision making based on the both the benefits, risks, and limitations of different tests, as well as personal, cultural and financial considerations.

**The impact of insurance coverage**

In assessing the challenges CNMs have faced as they begin incorporating NIPT into their offerings, we found many respondents cited difficulties understanding insurance reimbursement policies and navigating the process to providing this testing to patients who were underinsured or uninsured. Issues with insurance coverage and the cost of testing was also one of the most common reasons that CNMs reported not offering NIPT to their patients:

“I work in a clinic that is predominantly comprised of uninsured patients. I'm very limited in the testing that we can offer.”

“Insurance doesn't cover it for low-risk women and some of our patients have received big bills when they proceeded as low risk moms, to have the test”

As expected, the types of health insurance held by their patients influenced offering practices. CNMs caring for patients primarily insured through Medicaid were less likely to offer NIPT than
those caring for patients primarily insured through private plans. The shift toward offering NIPT to a wider variety of women, including those at low-risk, may result in an increasing number of situations for which insurance policies cover NIPT. In fact, the ACMG calls for “laboratories work with public health officials, policymakers, and private payers to make NIPS, including the pre- and post-test education and counseling, accessible to all pregnant women” (Gregg et al., 2016). If further research continues to suggest sensitivity and specificity are similar between the high- and low-risk obstetric populations, changes in insurance policies may increase the number of CNMs offering NIPT to their patients, alter the populations to whom they offer this testing, and increase patient uptake rates.

**Opportunities for further education**

We found that respondents desired further education regarding NIPT in four main areas: i) understanding positive results and indeterminate findings (including incidental findings), ii) the science and technology behind NIPT and how this is advancing, iii) practical/logistical considerations including information regarding insurance coverage and comparisons between the test offerings of different laboratories, and iv) the accuracy, reliability and positive predictive value of NIPT in various risk groups. These categories were consistent with the genetic counseling topics in which CNMs had the lowest levels of confidence: discussing care and management for a child with Down syndrome or one of the other conditions tested for (implications of a positive result) and discussing incidental/indeterminate findings. These perceived gaps in knowledge and areas of interest are important to consider when educating for CNMs and other providers about NIPT.
When asked about which resources were most helpful to learn about NIPT, about twice as many CNMs found those produced by testing companies (educational talks and/or literature/brochures) most helpful compared to those not provided by the testing laboratory (69% versus 29%). Furthermore, during pre-test counseling discussions with patients, 47% provide handouts produced by a lab company and 10% refer patients to a lab company website.

This widespread use of laboratory-produced sources to educate providers and patients about NIPT has both potential advantages and disadvantages. While they are plentiful and easily accessible, there is a possible conflict of interest when a laboratory testing company produces educational materials and stands to profit from increased use of their test. As ACOG states in their report on ethical issues in genetic testing, “Neutral counseling … may be compromised through the use of patient educational materials or counselors that are provided by a company that might profit from a patient's decision to undergo testing” (ACOG, 2008). At the same time, laboratory-employed genetic counselors are often involved in presenting information to healthcare professionals and writing or reviewing informational materials. A hallmark of the GC profession is providing information in a balanced way, including benefits, limitations, risks and alternatives of testing options (NSGC, 2006). Ideally, the increasing use of GCs in this role will encourage incorporation of balanced and accurate information in these resources and minimize potential bias despite this conflict of interest.

Another way private lab testing companies are involved in provider and client education is through their funding of independent organizations to disseminate genetic knowledge. For example Illumina and Sequenom, two large NIPT testing labs, have provided monetary support to help the Perinatal Quality Fund, a non-profit organization, develop their Genetic Education Module about NIPT for health care providers (Illumina, 2015; Sequenom, 2015). This is one
potential solution to increase provider and patient education while minimizing bias in educational materials. Through lab funding, independent organizations (either non-profits, educational institutions, or professional societies such as NSGC, ACOG, etc) are more likely to be able to produce potentially expensive educational talks, online courses, or print materials, ultimately allowing both parties to benefit from such a partnership.

Identifying the areas in which CNMs desire more education and those in which they have low perceived confidence provide opportunities to tailor education toward the most relevant and useful topics. We found that midwives who offer NIPT to their patients are significantly more confident discussing diagnostic testing options and the option of termination compared with those who do not offer this testing (p < 0.05). This may suggest that further education regarding the implications of positive results to the woman and fetus, rather than education about NIPT itself, may increase the probability that CNMs will offer this testing to their patients in the future.

**Study Limitations**

One limitation of this study was the low response rate (approximately 5%), which may raise questions regarding the generalizability of this sample to the population of CNMs providing prenatal care to women in the United States. Another limitation includes selection bias, since the recruitment email described the study as assessing how CNMs are integrating NIPT into their practice. It is possible those who do not offer this testing to patients were less likely to complete the survey, despite the email stating that CNMs were encouraged to participate whether or not they currently offer this testing. Also, levels of confidence were self-reported and do not necessarily represent actual levels of proficiency in discussing of the genetic counseling topics assessed. Finally, certified midwives (CM) were not eligible to participate in the study. CMs
have the same graduate training as CNMs and pass the same credentialing exam, but are not required to be a registered nurse (ACNM, 2014). Since CMs provide the same care to women as CNMs, and often work together with CNMs in the same practice, this study does not represent all advanced-degree midwifery practitioners offering NIPT to patients in the US.

**Future Research Recommendations**

This study provides initial insight into how CNMs are using NIPT in their practice, however, with the quickly changing technologies and evolving professional recommendations regarding prenatal testing options, up-to-date knowledge will require re-evaluating these practices on a continual basis. For example, the new 2016 ACOG/SFMF policy statement and ACMG committee opinion both support the option of offering NIPT to all pregnant women, not just those at high-risk for fetal aneuploidy. In response, CNMs’ practices may have changed, even within the short time between collection of responses and submission of this thesis. In addition, the short-answer responses we analyzed in this study highlight some of the challenges CNMs are facing as they begin offering NIPT to their patients, but qualitative studies, such as interviews or focus groups, would be helpful to further expand on these themes and maximize the relevance and usefulness of newly developed educational resources.
CONCLUSIONS

Our study found that the vast majority of CNMs are offering NIPT to at least some of their patients, but practices vary regarding whether this testing is offered to everyone or selectively to only a subset of patients. Like the constantly evolving professional guidelines regarding best practices for cell-free DNA screening in the general population, midwives have varied opinions regarding the value of this screening for patients at low risk for fetal aneuploidy, however, almost all offer this testing to patients at high risk. While there is a desire for further education about NIPT, the CNMs who do not offer NIPT attribute this fact to the structure of their practice or lack of insurance coverage, rather than low levels of confidence or understanding of various aspects of this testing. Respondents cited a desire for further education on a variety of topics, including the technology itself, conditions tested for, logistical concerns including insurance coverage, and understanding and counseling about positive and indeterminate results. Overall, this research provides a framework from which we can understand the current use of NIPT by CNMs, identifies challenges encountered since its integration into their practice, and suggests ways in which Genetic Counselors and the labs offering NIPT can best support CNMs’ efforts to broaden their scope of prenatal screening and navigate these challenges.
REFERENCES


APPENDICES

Appendix A: Survey

Q1 Thank you for your interest in this study! This research study is open to all Certified Nurse Midwives (CNMs) currently providing prenatal/antepartum care to women in the United States, whether or not you currently offer NIPT to your patients. The goal of this study is to investigate if and how CNMs are integrating non-invasive prenatal testing (NIPT) into their prenatal care. In particular, this study will investigate who is offered NIPT, explore CNMs’ experiences counseling patients about NIPT, and determine how CNMs are being educated about this testing. This anonymous survey is expected to take an average of 15 minutes to complete. Participation in this survey is completely voluntary, and you have the right to withdraw at any time or skip any questions without consequence. Your responses will be confidential, and no identifying information will be collected. Risks are minimal for involvement in this study, however, you may feel emotionally uneasy when asked certain questions. You may skip these questions, take a break from the survey, or withdraw from the survey at any time. There are no expected benefits to participants, but as a token of appreciation for your participation, you will have the opportunity to provide your email address at the end of the survey to be entered in a raffle for one of three $50 Amazon gift cards. If you choose to enter this raffle, your email address will not be linked to your survey responses. If you have questions or comments regarding this study, you may contact Lisa Weingarten (student researcher) at lweingarten@brandeis.edu or Dr. Judith Tsipis (faculty advisor) at tsipis@brandeis.edu. This study was reviewed and approved by the Brandeis University Institutional Review Board (IRB). If you have questions about your rights as a research subject please contact the Brandeis Institutional Review Board at irb@brandeis.edu or 781-736-8133. Solicitation of CNM/CM participants for this study has been approved by ACNM. By clicking the "Next" button, you consent to participation in this survey.

Q2 Do you currently provide prenatal (antepartum) care to women as a Certified Nurse Midwife (CNM)?
- Yes (1)
- No (2)

If No Is Selected, Then Skip To End of Survey

Q3 In which state do you primarily practice?
- I do not practice in the United States (1)
- Alabama (2)
- Alaska (51)
Arizona (3)
Arkansas (4)
California (5)
Colorado (6)
Connecticut (7)
Delaware (8)
District of Columbia (9)
Florida (10)
Georgia (11)
Hawaii (52)
Idaho (12)
Illinois (13)
Indiana (14)
Iowa (15)
Kansas (16)
Kentucky (17)
Louisiana (18)
Maine (19)
Maryland (20)
Massachusetts (21)
Michigan (22)
Minnesota (23)
Mississippi (24)
Missouri (25)
Montana (26)
Nebraska (27)
Nevada (28)
New Hampshire (29)
New Jersey (30)
New Mexico (31)
New York (32)
North Carolina (33)
North Dakota (34)
Ohio (35)
Oklahoma (36)
Oregon (37)
Pennsylvania (38)
Rhode Island (39)
South Carolina (40)
South Dakota (41)
Tennessee (42)
Texas (43)
Utah (44)
Vermont (45)
Virginia (46)
Washington (47)
Q4 Please indicate which of the following individuals at your practice/institution also provides prenatal/antepartum care to women, if any. Check all that apply.

- Another CNM (1)
- Obstetrician (OB) (2)
- Maternal Fetal Medicine specialist (MFM) (3)
- Nurse Practitioner (NP) (4)
- Registered Nurse (RN) (5)
- Genetic counselor (GC) (6)
- Physician assistant (PA) (7)
- Other (please specify): ____________________ (8)
- None (9)

Q5 Non-invasive prenatal testing (NIPT) is being used increasingly throughout the country to assess the chance that a woman’s fetus has a chromosome abnormality. This screening test analyzes cell-free DNA from the placenta found in the woman’s blood to detect these chromosome differences in the pregnancy. This test is also known as cell free fetal DNA testing (cffDNA), cell free DNA testing (cfDNA), non-invasive prenatal diagnosis (NIPD), and non-invasive prenatal screening (NIPS). This testing is offered by a variety of commercial testing companies, and each company has its own test name, for example, Informaseq (LabCorps), Qnatal (Quest), Maternity21 (Sequenom), Harmony (Ariosa), Panorama (Natera), etc. Please indicate which genetic screening or diagnostic tests you OFFER to your patients, if any, and the proportion of patients to whom you offer this testing.

<table>
<thead>
<tr>
<th>Test</th>
<th>I offer this to ALL my patients (1)</th>
<th>I offer this to SOME of my patients (2)</th>
<th>I DO NOT offer this to my patients (3)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Chorionic Villi Sampling (CVS) (1)</td>
<td>○</td>
<td></td>
<td>○</td>
</tr>
<tr>
<td>Amniocentesis (2)</td>
<td>○</td>
<td></td>
<td>○</td>
</tr>
<tr>
<td>Maternal Serum Screening (first screen, sequential, integrated, or quad screen) (3)</td>
<td>○</td>
<td></td>
<td>○</td>
</tr>
<tr>
<td>NIPT (4)</td>
<td>○</td>
<td></td>
<td>○</td>
</tr>
<tr>
<td>Nuchal Translucency measurement, on 1st trimester ultrasound (5)</td>
<td>○</td>
<td></td>
<td>○</td>
</tr>
<tr>
<td>2nd trimester level II</td>
<td>○</td>
<td></td>
<td>○</td>
</tr>
</tbody>
</table>
ultrasound / fetal anatomy survey (6)
Other (please specify): (7)
Other (please specify): (8)

Q6 For the following questions, definitions of 'high risk for fetal aneuploidy' and 'low risk for fetal aneuploidy' are available by hovering over the colored term.

Answer If Please indicate which genetic screening or diagnostic tests you offer to your patients, if any, a... NIPT - I offer this to SOME of my patients Is Selected
Q7 Of the women ACOG defines as high risk for fetal aneuploidy, to whom do you offer NIPT?
❖ All high risk women (1)
❖ Only women at high risk who bring up this option themselves (2)
❖ Only women at high risk who have insurance that will cover this testing (3)
❖ I do not see high risk women (4)
❖ Other (please specify): (5) ______________________

If I do not see high risk women Is Selected, Then Skip To Do you ever offer NIPT to women ACOG ...

Q8 Of the women ACOG defines as high risk for fetal aneuploidy who are offered NIPT, what proportion elects to proceed with NIPT?
❖ None (1)
❖ Few (2)
❖ Some (3)
❖ Most (4)
❖ All (5)

If Non-invasive prenatal testing (NIPT) is being used increasingly throughout the country to assess the chance a woman's fetus has a chromosome abnormality. This screening test analyzes cell-fre... NIPT - I offer this to ALL my patients Is Selected
❖ I do not see high risk women (6)

Answer If Non-invasive prenatal testing (NIPT) is being used increasingly throughout the country to assess the chance that a woman's fetus has a chromosome abnormality. This screening test analyzes cell-fre... NIPT - I offer this to SOME of my patients Is Selected
Q9 Do you ever offer NIPT to women ACOG defines as low risk for fetal aneuploidy?
❖ Yes (1)
❖ I do see low risk women, but I do not offer NIPT to these women (2)
❖ I do not see low risk women (3)

Answer If Do you ever offer NIPT to women ACOG defines as low risk for fetal aneuploidy? Yes Is Selected
Q10 Of the women ACOG defines as low risk for fetal aneuploidy, to whom do you offer NIPT?
- All low risk women (1)
- Only women at low risk who bring up this option themselves (2)
- Only women at low risk who have insurance that will cover this testing (3)
- Other (please specify): (5) ________________

Answer If Do you ever offer NIPT to women ACOG defines as low risk for fetal aneuploidy?
Yes Is Selected Or Non-invasive prenatal testing (NIPT) is being used increasingly throughout
the country to assess... NIPT - I offer this to ALL my patients Is Selected

Q11 Of the women ACOG defines as low risk for fetal aneuploidy who are offered NIPT, what
proportion elects to proceed with NIPT?
- None (1)
- Few (2)
- Some (3)
- Most (4)
- All (5)

If Non-invasive prenatal testing (NIPT) is being used increasingly throughout the country to
assess... NIPT - I offer this to ALL my patients Is Selected
- I do not see low risk women (6)

Q12 Based on your experiences, how often do the following factors play a role in a patient’s
decision to ELECT to have NIPT?

<table>
<thead>
<tr>
<th>Factor</th>
<th>Never (1)</th>
<th>Rarely (2)</th>
<th>Sometimes (3)</th>
<th>Often (4)</th>
<th>Always (5)</th>
</tr>
</thead>
<tbody>
<tr>
<td>No risk of miscarriage with NIPT (91)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Want as much information about pregnancy as possible (92)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Are considering termination if aneuploidy detected (93)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
| Want time to emotionally/financially prepare if having a baby with a
  chromosome abnormality (94)                                          |           |            |               |           |            |
<p>| NIPT is an accurate and reliable screen (95)                         |           |            |               |           |            |</p>
<table>
<thead>
<tr>
<th>Family/friends have done it (96)</th>
<th></th>
<th></th>
<th></th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Allows early determination of fetal sex (97)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Feel it is now a routine part of prenatal care (98)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Other (please specify): (99)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Q13 Based on your experiences, how often do the following factors play a role in a patient’s decision to DECLINE NIPT?

<table>
<thead>
<tr>
<th>Factor</th>
<th>Never (1)</th>
<th>Rarely (2)</th>
<th>Sometimes (3)</th>
<th>Often (4)</th>
<th>Always (5)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Do not want to know if the pregnancy has a chromosome aneuploidy (200)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Would rather just have diagnostic testing (CVS or amniocentesis) (201)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Would rather have another form of screening (maternal serum screening, ultrasound) (202)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Not concerned about their risk of chromosome aneuploidy</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
There is no cure for the chromosomal aneuploidies for which NIPT screens.

NIPT will provoke too much anxiety.

Not everyone has insurance that fully covers this testing.

Have ethical and/or religious objections to prenatal testing.

Other (please specify): 

Q14 How often do you provide your patients with a pre-test counseling discussion about NIPT?
- Never (1)
- Rarely (2)
- Sometimes (3)
- Often (4)
- Always (5)

If Never Is Selected, Then Skip To Who in your practice provides a pre-t...

Q15 Who else in your practice provides a pre-test counseling discussion about NIPT? Select all that apply.
- Another CNM (1)
- Obstetrician (OB) (2)
- Maternal Fetal Medicine specialist (MFM) (3)
- Nurse Practitioner (NP) (4)
- Registered Nurse (RN) (5)
- Genetic counselor (6)
- Physician assistant (PA) (7)
- Other (please specify): (8) ____________________
Q16 Please indicate which five (5) of the following are most important to include in a pre-test counseling discussion about NIPT.

- A description of Down syndrome (Trisomy 21) (1)
- A description of conditions other than Down syndrome (Trisomy 21) which are tested for (Trisomy 13, Trisomy 18, sex chromosome aneuploidies, etc.) (2)
- NIPT is only a screen; it is not diagnostic (false positive and false negative results can occur) (3)
- CVS or amniocentesis is needed to confirm a positive NIPT result (4)
- NIPT has better detection rates and lower false positive rates than maternal serum screening (5)
- NIPT does not screen for open neural tube defects (6)
- NIPT can determine the sex of the fetus (7)
- There is a possibility of a no-call or indeterminate test result with NIPT (8)
- There is a possibility of an incidental finding (e.g., maternal chromosome condition, maternal malignancy) with NIPT (9)
- NIPT has no risk of miscarriage (10)
- NIPT cannot screen for all chromosome abnormalities or genetic conditions (11)
- An explanation of how NIPT works (12)
- Cost and insurance coverage of NIPT (13)
- Other (please specify): ____________________

Q17 What resources, if any, do you provide to patients during your pre-test discussion of NIPT? Select all that apply.

- Printed material (brochure or handouts) - not provided by the testing laboratory (1)
- Testing laboratory’s printed material (brochure or handouts) (2)
- Testing laboratory’s website (3)
- Other website (4)
- Materials/handouts created by professional organizations (NSGC, ISPD, ACOG) (5)
- Materials (handouts, drawings, pictures) you created yourself (6)
- Other (please specify): ____________________
- None (7)

Answer: If How often do you provide your patients with a pre-test counseling discussion about NIPT? Never Is Selected

Q18 Who in your practice provides a pre-test counseling discussion about NIPT in your practice? Select all that apply.

- Another CNM (1)
- Obstetrician (OB) (2)
- Maternal Fetal Medicine specialist (MFM) (3)
- Nurse Practitioner (NP) (4)
- Registered Nurse (RN) (5)
- Genetic counselor (6)
- Physician assistant (PA) (7)
☐ Other (please specify): (8) ____________________
☐ None (9)

Q19 Have any of your patients received a positive/high-risk screen result on NIPT?
☐ Yes (1)
☐ No (2)
☐ Unsure (3)

Answer If Have any of your patients received a positive/high-risk screen result on NIPT? Yes Is Selected

Q20 For which condition(s) did this/these patients screen positive? Select all that apply.
☐ Trisomy 21 (Down Syndrome) (1)
☐ Trisomy 18 (2)
☐ Trisomy 13 (3)
☐ Sex chromosome abnormality (4)
☐ Microdeletion (5)
☐ Other (please specify): (6) ____________________

Answer If Have any of your patients received a positive/high-risk screen result on NIPT? Yes Is Selected

Q21 Did you provide post-test counseling to this/these patients?
☐ Yes (1)
☐ No (2)

Answer If Did you provide post-test counseling to this/these patients? No Is Selected

Q22 Who provided post-test counseling to this/these patients (from within or outside of your practice)? Select all that apply.
☐ Another CNM (1)
☐ Obstetrician (OB) (2)
☐ Maternal Fetal Medicine specialist (MFM) (3)
☐ Nurse Practitioner (NP) (4)
☐ Registered Nurse (RN) (5)
☐ Genetic counselor (6)
☐ Physician assistant (PA) (7)
☐ Other (please specify): (8) ____________________
☐ None (9)

Q23 Please indicate WHY you do not currently offer NIPT to any of your patients. Select all that apply.
☐ I do not feel comfortable explaining this testing (1)
☐ I do not see patients for whom NIPT is indicated/eligible for NIPT (2)
☐ I do not feel comfortable interpreting or disclosing results (3)
☐ This testing not always covered by insurance (4)
☐ Other providers in my practice/institution offer this test (5)
☐ This testing is not offered at my practice by ANY provider (6)
☐ There are no ACNM guidelines regarding NIPT (7)
I do not support prenatal genetic testing (8)
☐ Other (please specify): ____________________

Q24 Please include any additional comments below.

Q25 Do you plan to offer NIPT to at least some of your patients in the future?
☐ Yes (1)
☐ No (2)
☐ Unsure (3)
If No Is Selected, Then Skip To End of Block

Answer If Do you plan to offer NIPT to at least some of your patients in the future? Yes Is Selected Or Do you plan to offer NIPT to at least some of your patients in the future? Unsure Is Selected

Q26 What future changes would increase the probability of your offering NIPT to at least some of your patients?

Q27 Have you ever referred one of your patients to a genetic counselor for counseling about NIPT?
☐ Yes (1)
☐ No (2)

Answer If Please indicate which of the following individuals at your practice/institution also provides pre... Genetic counselor (GC) Is Not Selected And Have you ever referred one of your patients to a genetic counselor for counseling about NIPT? No Is Selected

Q28 Do you know of a genetic counselor or genetic counseling department to which you could refer a patient?
☐ Yes (1)
☐ No (2)

Answer If Have you ever referred a patient to a genetic counselor Yes Is Selected

Q29 In which situations have you referred at least some patients to a genetic counselor for counseling about NIPT? Select all that apply.
☐ Patients 35 or over (advanced maternal age) (1)
☐ Patients with a family/medical history of genetic conditions or birth defects (2)
☐ Patients with abnormal ultrasound findings indicative of a chromosomal condition (3)
☐ Patients who receive an abnormal maternal serum screen (1st trimester, Quad, etc.) (4)
☐ Any patient who expresses interest in learning more about NIPT prior to testing (5)
If Non-invasive prenatal testing (NIPT) is being used increasingly throughout the country to assess... NIPT - I DO NOT offer this to my patients Is Not Selected
☐ Patients who receive a negative NIPT result (6)
If Non-invasive prenatal testing (NIPT) is being used increasingly throughout the country to assess... NIPT - I DO NOT offer this to my patients Is Not Selected
☐ Patients who receive a positive NIPT result (7)
If Non-invasive prenatal testing (NIPT) is being used increasingly throughout the country to assess... NIPT - I DO NOT offer this to my patients Is Not Selected
Patients who receive a no-call/indeterminate NIPT result (8)

If Non-invasive prenatal testing (NIPT) is being used increasingly throughout the country to assess... NIPT - I DO NOT offer this to my patients Is Not Selected

Patients who receive a possible incidental finding from NIPT (9)

Other (please specify): (10) ____________________

Q30 NIPT is a new prenatal test that has only been clinically available since 2011. Which THREE (3) sources of information have you found most useful to learn about NIPT?

- Formal education/CNM degree program (1)
- Platform presentations or sponsored events at the ACNM annual meeting (2)
- Consulting with peers or co-workers (other than a genetic counselor) (3)
- Consulting with a genetic counselor (4)
- From self-directed review of literature (5)
- From educational talks/seminars/webinars presented by a laboratory or testing company (6)
- From educational talks/seminars/webinars NOT presented by a laboratory testing company (7)
- Literature/brochures produced by a laboratory or testing company (8)
- Literature/brochures NOT produced by a laboratory or testing company (9)
- Resources provided by a professional organization (ACOG, ISPD, NSGC, etc.) (10)
- Other source (please specify): (11) ____________________
- I have not received useful information about NIPT (12)

Q31 Please rate your level of CONFIDENCE in discussing the following topics with patients, from 1 (not at all confident) to 5 (completely confident).

<table>
<thead>
<tr>
<th>Topic</th>
<th>Not At All Confident (1)</th>
<th>Slightly Confident (2)</th>
<th>Somewhat Confident (3)</th>
<th>Mostly Confident (4)</th>
<th>Completely Confident (5)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Care and management for a child with Down syndrome/Trisomy 21 (1)</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>Care and management for a child with a chromosome condition OTHER than Down syndrome (2)</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>Diagnostic testing options to confirm screening results (CVS or amniocentesis) (3)</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>If Non-invasive prenatal testing (NIPT) is being used increasingly throughout the country to assess...</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>NIPT - I DO NOT offer this to my patients Is Not Selected</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>The accuracy and reliability of NIPT (4)</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>If Non-invasive prenatal testing (NIPT) is being used increasingly throughout the country to assess...</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>NIPT - I DO NOT offer this to my patients Is Not Selected</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Implications of a no-call/indeterminate result on NIPT (5)</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>If Non-invasive prenatal testing (NIPT) is being used increasingly throughout the country to assess...</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>NIPT - I DO NOT offer this to my patients Is Not Selected</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Potential for incidental findings from NIPT (6)</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Facilitating decision making regarding genetic testing options (7)</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Counseling</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
</tbody>
</table>
regarding the option to end a pregnancy after a positive (abnormal) genetic testing result (8)

Answer If Non-invasive prenatal testing (NIPT) is being used increasingly throughout the country to assess... NIPT - I DO NOT offer this to my patients Is Not Selected

Q32 Please describe any thoughts that you have regarding the value of NIPT for your patients.

Q33 If you were to attend one lecture regarding NIPT, what would you like to learn more about?

Q34 How many years have you have been practicing as a certified nurse midwife (CNM)?

Q35 What is your age?

Q36 What is your gender?
- Male (1)
- Female (2)
- Other (3)

Q37 Which best describes your primary employer?
- Physician-owned practice (1)
- Midwifery-owned practice (2)
- Hospital/medical center (3)
- Educational institution (4)
- Community Health Center (5)
- Military (6)
- Other (please specify): (7)

Q38 On average, how many prenatal patients do YOU see each WEEK?
- 1-5 (1)
- 6-10 (2)
- 11-15 (3)
- 16-20 (4)
- 21-25 (5)
- Over 25 (6)

Q39 Approximately what proportion of your patients has medical insurance coverage?
- 0-25% (1)
- 26-50% (2)
- 51-75% (3)
- 76-100% (4)
Q40 What is the PRIMARY payment method for the MAJORITY of your patients?
- Medicaid (1)
- Private insurance (2)
- Self payment (4)
- Other (please specify): (5) ____________________

Q41 Congratulations, you have reached the end of the survey. Please click NEXT to submit your responses.

Raffle

Q1 Thank you for your participation! Your responses have been recorded. If you would like to be entered in the raffle for one of 3 $50 Amazon gift cards, please enter your email address below. This information WILL NOT be linked in any way to your survey data.
Appendix B: Recruitment Email

Dear ACNM member,

Do you provide prenatal/antepartum care to women as a Certified Nurse Midwife (CNM)?

If so, I invite you to participate in a research study investigating if and how CNMs are integrating non-invasive prenatal testing (NIPT) into their prenatal care. The primary goals of this research study are to investigate who is offered NIPT, explore CNMs' experiences counseling patients about this testing, and determine how CNMs learn about this testing themselves. **This study is open to all CNMs who provide prenatal care in the United States, whether or not you currently offer NIPT to your patients.**

This research study is being carried out as a part of my Master's thesis for Brandeis University's Genetic Counseling Program. Solicitation of CNM/CM participants for this study has been approved by ACNM.

This anonymous survey is expected to take an average of 15 minutes to complete. Upon completion of the survey, you will have the opportunity to enter a raffle to win **one of three $50 Amazon Gift Cards.** If you choose to enter the raffle, your survey responses will not be connected to your email address.

Please click [here](#) to take the survey.

Please consider forwarding this email to any CNM colleagues who may be interested in participating in the survey.

If you have questions or comments regarding this study, feel free to contact me at [lweingarten@brandeis.edu](mailto:lweingarten@brandeis.edu), or Judith Tsipis (faculty advisor) at [tsipis@brandeis.edu](mailto:tsipis@brandeis.edu). Any questions regarding your rights as a research participant can be directed to the Brandeis IRB at [irb@brandeis.edu](mailto:irb@brandeis.edu) or 781-736-8133.

Thank you in advance for your participation!

Sincerely,

**Lisa Weingarten, MS**  
Genetic Counseling Master's Candidate  
Class of 2016  
Brandeis University

**Judith Tsipis, PhD**  
Director, Genetic Counseling Program  
Professor of Biology  
Brandeis University