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Recall Of Informed Consent For Prenatal Aneuploidy Screening

Taylor Cain
Sarah Lawrence College

Michelle Kao
Sarah Lawrence College

Elena Cothalis
Sarah Lawrence College

Pranali Shingala
Sarah Lawrence College

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Taylor Cain[1], Michelle Kao[1], Elena Cothalis[1], Pranali Shingala[1], Katie Stoll[2], Laura Hercher[1]

[1]Joan H. Marks Graduate Program in Human Genetics, Sarah Lawrence College, Bronxville, New York
[2] Genetic Support Foundation, Olympia, Washington

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Abstract

Several forms of prenatal screening and diagnostic testing are available that can provide information about the likelihood of a genetic or chromosomal condition in pregnancy. Each of the available technologies entail unique benefits and limitations, and patient comprehension of the differences among these tests is crucial to uphold the principle of informed consent. The primary research goal of our study was to establish what women recall of the benefits, risks, and limitations of the prenatal aneuploidy screening they were offered as a part of their prenatal care by their medical provider. A total of 349 women were surveyed and 182 met eligibility criteria having had a recent or third-trimester pregnancy. Overall knowledge scores for participants ranged from 0 to 82.9 out of 100, with an average score of 32.8 (SD = 21.9). We found significantly higher scores in women who were offered testing by genetic professionals, met with a genetic counselor in prior pregnancies, were 35-years or older, or were given the choice of both screening and diagnostic tests. Our data support the importance of a thorough informed consent when discussing prenatal aneuploidy testing with patients.

Key words: Prenatal aneuploidy screening, Informed consent, Patient education, Patient knowledge, Prenatal diagnostic tests

Introduction

According to practice guidelines from the American College of Obstetricians & Gynecologists (ACOG), prenatal testing should be made available to all women who wish to determine the risks of having a child with certain birth defects or genetic conditions (Rose & Mercer, 2016). Aneuploidy, or the condition of having more or less than the normal complement

of chromosomal material, is among the most commonly tested-for conditions. Several forms of prenatal screening and diagnostic testing are available that detect aneuploidy or the risk of aneuploidy, and each of these technologies has unique benefits and limitations. Because of the complexities involved with prenatal testing, several organizations, including ACOG, the American College of Medical Genetics and Genomics (ACMG), and the National Society of Genetic Counselors (NSGC), have published guidelines regarding testing for aneuploidy in pregnancy. All of these professional guidelines stress the importance of informed consent prior to initiating testing (Gregg et al., 2016; Rose & Mercer, 2016; Wilson et al., 2013). This includes a discussion of the risks, benefits, and limitations of each test, as well as a consideration of the patient's clinical circumstances, values, and preferences. However, it is not clear to what extent this requirement for informed decision-making is being met in practice.

Analyte screening, ultrasound, and non-invasive prenatal screening (NIPS) are among the tools used to screen for aneuploidy, and a major point of comparison between these modalities has been the clinical validity of the screening tests. Since its introduction in 2011, NIPS has been shown to have the highest detection rate for Down syndrome and lowest false positive rate for traditional aneuploidies relative to other screening methodologies (Pergament et al., 2014; Taneja et al., 2016). Despite this advantage, NIPS is not the most effective test for all purposes. For example, NIPS does not assess the risk of neural tube defects and overall may be less likely to detect other genetic and chromosomal conditions than analyte screening (Baer et al., 2015; Norton, Jelliffe-Pawlowski, & Currier, 2014; Shaffer & Norton, 2018). Given that common aneuploidies make up a lower fraction of the total risk in younger women, there has been some suggestion that analyte screening is more likely to lead to diagnosis of a clinically significant condition than NIPS in many women. Similarly, fetal ultrasound is useful in detecting structural

anomalies and soft markers that may indicate an increased risk for aneuploidy as well as other genetic conditions.

Diagnostic tests are also available in the form of chorionic villi sampling (CVS) and amniocentesis which determine, with as much certainty as possible, whether the fetus is aneuploid. Both diagnostic tests allow for further genetic studies beyond those for aneuploidy, including, but not limited to, single gene testing and chromosomal microarray. CVS allows for early diagnosis of the fetus by testing cells directly from the placenta. However, in 2% of viable pregnancies, chromosomal abnormalities exist and are confined to the placenta, which may result in the misdiagnosis of an unaffected fetus (Kalousek & Vekemans, 1996). Amniocentesis remains the gold standard for diagnosing aneuploidy, as it directly tests fetal cells in the amniotic fluid. Other advantages of amniocentesis include the testing for neural tube defects, and fetal infections. Due to the invasive nature of both procedures, they carry a small risk of miscarriage due to infections or amniotic sac rupture, generally quoted as less than 1% (Choudry, Masood, & Ahmed, 2012; Eddleman et al., 2006; Tabor et al., 1988).

Despite the emphasis on informed consent prior to prenatal aneuploidy testing, not all patients receive adequate counseling. Many factors have been found to influence uptake of aneuploidy screening including education level, income, experience with genetic testing, willingness to consider abortion, and experience with disability (Sayres, Allyse, Goodspeed, & Cho, 2014). Women also report considering multiple factors when deciding if and how to pursue prenatal aneuploidy testing including accuracy, safety, timing, and ease of testing (Lewis, Hill, & Chitty, 2016). Concerns have been raised regarding the impact of routinization on patient choice in initiation of testing (Allyse, Sayres, Goodspeed, & Cho, 2014). Moreover, not all women who are informed about the details of prenatal aneuploidy tests demonstrate complete knowledge of

the risks, benefits, and limitations of testing (Rachel Farrell, Hawkins, Barragan, Hudgins, & Taylor, 2015; Lewis, Hill, Skirton, & Chitty, 2016; Piechan et al., 2016). Informed consent may also be limited by provider knowledge and attitudes. Some general practitioners and obstetricians have indicated that they would offer termination of a pregnancy following positive NIPS, suggesting that not all providers understand the limitations of the test (Chan, Johnson, Wilson, & Metcalfe, 2018). Likewise, some maternal-fetal-medicine fellows have indicated that they are uncomfortable ordering aneuploidy screening (Swaney, Hardisty, Sayres, Wiegand, & Vora, 2015). Furthermore, studies examining obstetricians' experiences with prenatal aneuploidy screening have revealed that providers struggle with the limited time available to adequately counsel patients about options for prenatal screening, and that not all obstetricians provide pre-test counseling or refer patients to genetics (R. M. Farrell, Agatisa, Mercer, Mitchum, & Coleridge, 2016; Gammon, Kraft, Michie, & Allyse, 2016).

Purpose of the Study

Only a few studies to date have examined the informed consent process for women undergoing routine aneuploidy screening from an obstetrician or other non-genetics medical provider. We developed a survey to gather information regarding what women recall of the informed consent process and the benefits, risks, and limitations of prenatal aneuploidy screening when offered as a part of their prenatal care by an obstetrician or other non-genetics medical provider. The purpose of this study was to assess the effectiveness of current practice in helping women make informed decisions about prenatal screening.

Methods

Participants and Procedures

Ethical approval was obtained from the Sarah Lawrence College Institutional Review Board. To avoid institutional bias, participants were recruited through social media including Facebook groups, online forums, and Twitter for mothers and not through providers or health care networks. For the purpose of the study, only women who were in their third trimester of pregnancy or had given birth within 12 months of completing the survey were included in the analysis. All data were collected between December 2018 and February 2019.

Instrumentation

An online survey was administered through SurveyMonkey to obtain information regarding women's experiences with prenatal aneuploidy testing offered to them in their current or most recent pregnancy. At the start of the survey, participants were informed that the study was voluntary and they could discontinue the survey at any time. The survey primarily consisted of multiple-choice questions with one free-response question. The survey was grouped into three categories that assessed demographics, prenatal care experiences, and knowledge of prenatal aneuploidy testing.

In the demographic section, participants were asked to provide information regarding their age at birth at the time of their current or most recent pregnancy, ethnicity, annual household income, primary language spoken at home, and education level. The prenatal care section consisted of questions regarding whether the participant had any children prior to the study, if the participant had seen a prenatal genetic counselor during this or any past pregnancy, the type of provider (if any) that discussed prenatal aneuploidy testing with them, the types of prenatal tests that were offered, time spent discussing the tests, and prenatal test decision-making. A free-response question asked for any additional comments regarding their experience.

The final section consisted of eight multiple-response questions that tested participants' knowledge of the benefits and limitations of prenatal screening, diagnostic testing, and ultrasound. Participants were asked which of the prenatal tests could provide specific information on the fetus, such as the ability to detect aneuploidy, to diagnose aneuploidy, or to guarantee a healthy baby. There were 8 possible answer choices: First-trimester serum screen, quad serum screen, NIPS, ultrasound, amniocentesis, CVS, none of these, and I don't know. To create a knowledge score, one point was awarded for each correct answer chosen, one point was deducted for each incorrect answer chosen, and zero points were awarded or deducted if the choice was left blank. Participants that received negative points were adjusted to zero. Because each quiz question had a different number of correct answers, the questions were normalized so that each question held equal weight in the final score. This was done by dividing the participants total points by the number of correct answers for each question and multiplying this value by 10. The scores for each question were then summed, divided by the 80 total points, and multiplied by 100 to provide the participant's knowledge score.

Participants were also given the option to provide contact information if they wished to enter a raffle for one of five $100 Amazon gift cards. The survey was piloted with one eligible participant whose response was not included in the data analysis. Based on feedback from the pilot, the wording of the survey was revised to minimize confusion.

Data Analysis

All data was de-identified at the start of analysis. A total of 349 survey responses were received. Of those responses, 167 were either incomplete or did not meet eligibility criteria based on the timing of their pregnancy, resulting in a total of 182 eligible responses. For the remaining

182 responses, knowledge scores were calculated and survey data was coded. All statistical analyses were performed using SPSS.

One-way ANOVAs were performed to compare knowledge scores to the amount of education received (0-5 mins, 5-15 mins, 15+ mins) and types of testing offered (screening, diagnostic, both, none). Independent t-tests were performed to compare knowledge scores with advanced maternal age (AMA) status, previous children prior to the study, prior experience with a prenatal genetic counselor, provider type (genetic, non-genetic), and how recently the participant saw a provider based on eligibility criteria (currently pregnant, given birth). For all statistical analyses, a significance threshold was set at $p < 0.05$.

In addition to statistical analyses, a qualitative analysis was performed to evaluate an open-ended question that asked about further comments relating to participants' prenatal genetic testing experience. Of the 182 participants, 42 women provided a response in this section. Four project investigators independently reviewed these responses, and common themes were identified and discussed. This process generated a list of common themes. The project investigators then individually coded participants' responses. The team compared the coded responses, discussed discrepancies, and developed rules to make coding consistent, after which the codes were finalized. Disagreements were addressed through conversations until the coders reached consensus. A consensus was met when a minimum of three of the four coders investigators were in agreement. Using the final codes, percentages for each theme were calculated.

Results

The study had 182 eligible participants. Participant demographics are summarized in Table I. A majority of participants identified as Caucasian or white (61.5%, n=112), used English

as a primary language (92.9%, n=169), had a family household income of at least $75,000 (68.1%, n=124), and had a college or university degree or higher (85.2%, n=155). At the time of surveying, 45 participants were in the third trimester (24.7%) while 137 participants had given birth within the last 12 months (75.3%). Participants ranged between 18-47 years old, with an average age of 32.5 years.

Information regarding participants' prenatal history can be found in Table II. Of the 174 women who reported their age, the majority of participants were under the age of 35 (65.7%, n=115). Similarly, of the 182 eligible participants, most did not have any prior children (52.7%, n=96). Some participants noted that they had met with a prenatal genetic counselor at some point in the past, prior to their current or most recent pregnancy (31.3%, n=57). Most participants noted that the primary person providing information to them about prenatal genetic testing for their current pregnancy was an obstetrician (56.6%, n=103). Other providers discussing prenatal testing options included a midwife (13.7%, n=25), genetic counselor (9.9%, n=18), and physician's assistant or nurse (7.1%, n=13). When asked about how much time providers spent educating participants about prenatal aneuploidy tests, the most common answers were "between 5 and 15 minutes" (48.9%, n=89), followed by "less than two minutes" (30.2%, n=55), and "fifteen to thirty minutes" (11.0%, n=20). Of note, 39 participants (35%) who reported obstetrician as their main provider (n= 103) spent less than two minutes in this discussion.

A summary of the topics discussed and prenatal aneuploidy tests offered by the providers is in Table III. A majority of women recall that they were offered first-trimester serum screen (58.2%, n=106) and non-invasive prenatal screen (58.8%, n=107). Only a small portion of the participants report that they were offered diagnostic procedures (amniocentesis, 17.0%, n=31; CVS, 11.5%, n=21). However, 13.2% of women don't recall what they were offered (n=24).

Participants indicated that, when describing testing, most providers addressed what the test can detect (86.2%, n=150) but less than half addressed what the test cannot detect (44.8%, n=78). Less than half of the women reported that methodology (47.7%, n=83), detection rates (40.8%, n=71), or physical risks to the pregnancy (37.9%, n=66) were discussed when the tests were offered. Additionally, women's testing preferences and personal beliefs were generally not addressed during the discussion (28.7%, n=50).

The participants' testing uptake and factors influencing their decisions are also summarized in Table III. Of the participants who were offered specific aneuploidy testing, most women report that they chose to have first-trimester screen (72.6%, n=77), quad serum screen (67.7%, n=42), and NIPS (78.5%, n=84). Women were less likely to report having diagnostic procedures, such as amniocentesis (25.8%, n=8) or CVS (9.5%, n=2). Approximately one in seven participants declined testing (14.4%, n=25) and approximately one in nine couldn't recall the type of testing they had chosen (11.5%, n=20).

Of the 174 women that were offered any aneuploidy testing, most of the participants indicated that their desire to learn more about the health of their baby influenced their decision on testing (67.8%, n=118). Less commonly, costs (25.9%, n=45), physical risks (24.1%, n=42) , and personal beliefs about testing (16.1%, n=28) were considered. The ability to identify the sex of the fetus influenced testing uptake in 20.7% of the women (n=36). A third of the participants pursued testing because their provider recommended it (32.2%, n=56).

Participants' responses to individual quiz questions can be found in Table IV. For most questions, less than half of the participants answered the question correctly. Only for three questions did over half of the participants select a correct answer: that "ultrasound can detect the sex of the baby" (72.5%, n=132), that "there is a risk for miscarriage associated with

amniocentesis" (70.3%, n=128), and that "no test is able to determine that a baby will be born healthy" (64.3%, n=117).

Overall knowledge scores for participants ranged from 0 to 82.9, with an average score of 32.8 (SD = 21.9). The relationship between knowledge scores and a variety of participant variables can be found in Table V. Participants that were or would be at least 35 years old at the time of their delivery had significantly higher knowledge scores compared to younger women ($p<0.001$). Similarly, women with previous pregnancies had significantly higher knowledge scores compared to women who were pregnant or had given birth for the first time ($p=0.006$). Provider type also influenced knowledge scores. Participants that had met with a prenatal genetic counselor during previous pregnancies, and participants that met with a genetics specialist to discuss prenatal aneuploidy testing for their current or recent pregnancy, had significantly higher knowledge scores compared to those who did not, or those that met with a non-genetics specialist, respectively ($p<0.001$, $p=0.008$). Women who reported getting more time to discuss aneuploidy tests with providers tended to have higher knowledge scores, but this was not statistically significant ($p=0.056$). There was also a significant difference in knowledge scores related to the type of testing that was offered according to their reports, with women offered both screening and diagnostic testing scoring the highest ($p=0.011$). There was no significant difference in scores between women who were currently pregnant and those that gave birth within the past 12 months ($p=0.919$).

Of the 182 participants that were eligible for the study, 42 women provided additional comments about their prenatal testing experience. Common themes in participants' open-ended responses can be found in Table VI. The most common themes reflected experiences with providers. Some participants expressed a positive experience (14.3%, n=6), but more participants

expressed a negative experience with providers (38.1%, n=16). Negative experiences with providers were further broken down into specific provider limitations including: incomplete patient education (19.0%, n=8), absence of informed consent (9.5%, n=4), directive providers (7.1%, n=3), and lack of provider knowledge (7.1%, n=3).

In addition, participants also addressed specific factors influencing uptake of testing including insurance coverage and cost of testing (28.6%, n=12) and limitations of testing (21.4%, n=9). Other comments indicated specific participant characteristics that respondents identified as relevant in their choice of testing, most commonly being information-seeking (26.2%, n=11) or deliberative (14.3%, n=6). Under the theme of 'experience with testing', participants were equally likely to report a positive experience (9.5%, n=4) as they were to report a negative experience with testing (9.5%, n=4). Additionally, an identical number of participants indicated that testing was not necessary (9.5%, n=4).

Discussion

As prenatal aneuploidy testing is becoming routine in the clinical setting, there is an increasing need to assess women's understanding of the benefits, risks, and limitations of aneuploidy testing. To our knowledge, no study to date has examined the informed consent process for women undergoing routine aneuploidy testing from an obstetrician or other non-genetics medical provider. This study offers an understanding of what women recall from their prenatal aneuploidy testing experience and helps assess the effectiveness of current practice in helping women make informed decisions about prenatal testing.

Summary of findings

Our study suggests that most women are limited in their understanding of the details of prenatal aneuploidy testing, as evidenced by low knowledge scores. The participants' average

knowledge score was 32.8 out of 100 (SD = 21.9). With the exception of three answers, less than half of the participants selected the correct response for each question (Table IV). In addition, for each question, multiple women indicated that they did not know the answer. This suggests that women making reproductive health decisions are inadequately informed about prenatal aneuploidy testing. Our findings support previous studies showing that women undergo testing with a limited understanding of why they are being tested, what they are being tested for, and what the tests can tell them (Johnston, Farrell, & Parens, 2017; Parham, Michie, & Allyse, 2017).

Participants' poor knowledge scores are likely associated with limitations in the prenatal informed consent process. Professional organizations, including ACOG, recommend that all women are offered the option of prenatal aneuploidy testing. Despite this recommendation, 4.4% of women reported they were not offered prenatal aneuploidy testing (Rose & Mercer, 2016). While a small percentage, this is not an insignificant number if applied to the general population of pregnant women. Moreover, a major challenge of the informed consent process is providing women with enough information to make an informed decision on which testing, if any, is right for them. This study indicates that providers do not always spend adequate amounts of time discussing prenatal aneuploidy testing. Over 1/3 (n=67) of participants reported that providers spent a maximum of five minutes discussing aneuploidy testing. More specifically, 39 participants recalled that their obstetrician spent less than two minutes in this discussion, of particular significance since the obstetrician is the most frequent provider of information on aneuploidy testing (n=103) reported in this study. It is difficult to comprehend how women can receive adequate information and exercise informed consent for a complex decision in such a short amount of time.

Furthermore, a majority of women reported that providers did not address what the tests can't detect, testing methodology, detection rates, or physical risks to the pregnancy. This lack of discussion was reiterated in close to 1/5 of participants' open-ended responses. This includes the comments that "most of what I marked in previous questions as 'provider discussed with me' was due to my and my husband's questions", "I wish things had been explained more clear regarding dangers and what the tests actually entailed", and "I wish I was given more information on the topic from the doctor rather than having to read about it". Allotting sufficient time for a comprehensive discussion on testing may increase women's knowledge of testing.

The participants who met with a non-genetics provider had a significantly lower knowledge score compared to the participants who met with a genetics specialist. The inadequate patient understanding about prenatal screening and testing options may be due in part to lack of provider knowledge about prenatal genetic screening. Findings concluded that 7.1% of participants felt their providers lacked knowledge. This thought was echoed by one participant who stated that "The midwife I spoke to was very poorly informed." Non-genetic providers may offer screening options during patient visits, but the lack of in-depth discussion or knowledge on the providers side raises the issue that patients may not be receiving adequate knowledge needed for informed consent. In previous studies on providers, 45% of obstetricians indicated that their residency training in this prenatal counseling was inadequate to nonexistent, and a third of the physicians use information provided by commercial laboratories as initial source to acquire knowledge about NIPT (Cleary-Goldman et al., 2006; R. M. Farrell et al., 2016). Genetic providers are well-versed in testing details, and prior studies have shown that genetic counseling was found to positively correlate with sufficient knowledge regarding genetic testing (Sheinis, Bensimon, & Selk, 2017).

In this study, some maternal factors were determined to significantly influence women's knowledge in prenatal testing, including maternal age and number of children. Maternal age plays a significant role in prenatal care as women who are 35 and older are considered high-risk for having a child with aneuploidy (Driscoll & Gross, 2009). Although individual institutions differ in practice, these women are more likely to receive additional care, such as a referral to meet with a genetic counselor, additional ultrasound and obstetric monitoring, and screening and diagnostic tests for aneuploidy (Johnson et al., 2012). The high-risk status and the differential treatment from healthcare providers may result in their higher knowledge scores compared to women who are younger with lower risks. Additionally, women who have had previous pregnancies prior to this study, scored higher than first-time mothers. This suggests that women who have been pregnant in the past may benefit from the reiteration of the prenatal care process.

Another focus of our study was to investigate participants' choices in prenatal aneuploidy testing. More than half of participants indicated that their desire to learn more about the health of their baby influenced their decision on testing. This result suggests that the health of the baby is a driving factor for women when making a decision about prenatal testing. Other less factors influencing participants' decision on testing include costs: risks associated with testing, limitations of testing, and the desire to learn more about the sex of the baby. Not all women valued the same information, and this highlights the need for a complete informed consent. Nevertheless, close to 1 in 10 participants that responded to the open-ended question indicated that they did not receive informed consent for testing. Women expressed "The test was being performed without asking me", "I think I had the serum screening but it was never really discussed with me so I'm not 100% sure", and "I asked when I would be having the testing for Down syndrome... It was noted to me that it already had been performed, and all was normal.

Therefore, I do not recall how the genetic testing was done". These results are consistent with studies that found that obstetrical providers are less likely to believe that informed consent should be obtained prior to NIPS (Ruth Farrell, Mercer, Agatisa, Smith, & Philipson, 2014; Silcock, Liao, Hill, & Chitty, 2015; van den Heuvel et al., 2010).

Another component of informed consent is considering patient preferences for testing; yet, a vast majority of providers did not discuss beliefs and preferences about whether or not testing was right for the patients (71.3%, n=124). Similarly, close to ⅓ of women indicated that their decision to pursue prenatal aneuploidy testing was influenced by provider's recommendations, suggesting that some providers are directive in their counseling approach. This was further emphasized in the open-ended comments. One woman reflected that her medical providers "were pushing for amnio". Another woman commented that she "took the test because [her] provider provided [it] as a non-option test". These findings are consistent with a study that observed that physicians were less consistent in the practice of non-directiveness relative to genetic counselors, which was thought to be caused by an inherent bias in what they believe is best for their patients (Botkin, 1990). Together, these findings indicate that the requirement for informed decision-making is generally not being met in clinical practice. In the select cases where participants were given the opportunity to make an informed decision, participants' open-ended responses reflected positive experiences with providers: "I am very grateful to have been presented with options for different types of genetic testing, especially since there were some results which were unfavorable" and "I appreciate that my health care provider did not try to influence my decision to forego first trimester testing".

Study Limitations

As with many quantitative research surveys which rely on volunteer participants, the 182 eligible individuals who elected to participate in our study may not have been representative of all mothers; for example, they may have had a particular interest in prenatal screening. Our results could have also been skewed if participants were compelled to take the survey due to either an unusually positive or negative experience with our topic, which may also have influenced their survey responses. Our participant demographics also show a skew towards Caucasian women (61.5%) and women who are educated (45.2% completed at least a university or college degree). Furthermore, the internet was also used for the means of survey distribution, which allowed us to succeed in recruiting respondents from a broader geographic region. However, even so, our results overrepresent the east coast. This may have also marginalized mothers who are less inclined to use social media.

Research Recommendations

Our study was designed to investigate patient memory retention and understanding of prenatal testing options. The administration of a survey on this topic directly following an appointment with a provider may offer a clearer picture of what new and expectant mothers are being told about their screening options. In addition, further surveying of providers may provide a more complete understanding of patient education and informed consent in prenatal settings. Our study suggests a lack of informed consent in the realm of prenatal screening that patient education guidelines may help to close and further investigations in this area may be warranted.

Practice Implications

The diverse number of prenatal screening and testing options has changed the practice of prenatal care. The differences in test characteristics should be made clear to both provider and patients. A goal of genetic counseling is to support informed and autonomous decision making in

order to improve patient outcomes. Comprehensive training from non-genetic professionals in aspects of this practice along with the specifications of prenatal genetics may aid in informed decisions about prenatal testing from a patient standpoint. The implementation of standard consenting protocols and decision aids about screening and testing options may increase knowledge and satisfaction of both the patient and provider. Educational tools and patient-friendly consent forms could allow patients to understand the specifics of testing and enable them to make an informed decision about their pregnancy care.

Conclusions

During the course of their pregnancy, some women are confronted with a complex and ever-changing array of prenatal testing options. Striving for informed consent with prenatal testing improves the likelihood that patient decisions are made with sufficient knowledge and understanding, and are consistent with the patient's values and attitudes. However, our data indicates that, with no patient education guidelines in place, the quality and quantity of their informed consent process may vary greatly. This study also found that meeting with a trained genetic professional, AMA status, as well as having both screening and diagnostic options, maximize patient understanding and retention of genetic testing information. Though further analysis remains to be done on how to yield higher levels of patient knowledge, our study demonstrates that, under current practice, a significant percentage of women are not given the opportunity to make their own decisions in regards to their prenatal healthcare. As the prenatal testing choices available to women increase, ensuring healthcare providers are adept at educating women on the benefits and limitations of the myriad screening and diagnostic options available to them is critical.

In recent years, the rapid incorporation of aneuploidy testing in routine prenatal care has threatened the ability of women to make autonomous decisions. This emerging practice rests on the assumption that having prenatal tests is a woman's preferred choice, and therefore that offering them the opportunity to make a choice is unnecessary, thus undermining women's reproductive autonomy (Kater-Kuipers, de Beaufort, Galjaard, & Bunnik, 2018). As seen in this study, when given the choice, 14.4% of women did not have testing done. The issue of routinization and lack of informed consent may in turn, trivialize more difficult and controversial topics such as the choice of doing invasive testing, termination of pregnancy, and disability (Kater-Kuipers et al., 2018). As one of our participants stated, "just because we can do hundreds of tests does not mean that we should do them."

All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000 (5). Informed consent was obtained from all patients for being included in the study.

Conflict of Interests

Authors Taylor Cain, Elena Cothalis, Michelle Kao, and Pranali Shingala declare that they have no conflict of interest.

Acknowledgements

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Table I. Participant demographics

		n (%)
Ethnicity	African American or Black	5 (2.7%)
	Asian or Asian American	33 (18.1%)
	Caucasian or White	112 (61.5%)
	Hispanic or Latino	8 (4.4%)
	Jewish	3 (1.6%)
	Native American or Alaska Native	1 (0.5%)
	Mixed	20 (11.0%)
Primary Language	Chinese	9 (4.9%)
	English	169 (92.9%)
	Spanish	3 (1.6%)
	Yiddish	1 (0.5%)
Family Income	Under $15,000	4 (2.2%)
	Between $15,000 and $29,999	9 (4.9%)
	Between $30,000 and $49,999	14 (7.7%)
	Between $50,000 and $74,999	31 (17.0%)
	Between $75,000 and $99,999	36 (19.8%)
	Between $100,000 and $150,000	39 (21.4%)
	Over $150,000	49 (26.9%)
Education Level	Some high school	1 (0.5%)
	High school graduate, diploma, or equivalent	10 (5.5%)
	Technical or Associates degree	3 (1.6%)
	Some college	13 (7.1%)
	College or university degree	81 (44.5%)
	Graduate degree	74 (40.7%)

Table II. Participants' prenatal history

		n (%)
Advanced Maternal Age Status	<35 years	115 (65.7%)
	≥35 years	60 (34.3%)
Number of Children	0	96 (52.7%)
	1	62 (34.1%)
	2	16 (8.8%)
	3 or more	8 (4.4%)
Previously Met with a Prenatal Genetic Counselor	Yes	57 (31.3%)
	No	125 (68.7%)
Provider Type	Obstetrician	103 (56.6%)
	Midwife	25 (13.7%)
	Genetic counselor	18 (9.9%)
	Physician's assistant/nurse	13 (7.1%)
	Maternal-fetal medicine specialist	6 (3.3%)
	Family doctor/general practitioner	6 (3.3%)
	Fertility doctor	5 (2.7%)
	Other	5 (2.7%)
	Geneticist	1 (0.5%)
Time Spent Educating	No time	9 (4.9%)
	Less than 2 minutes	55 (30.2%)
	Between 2 and 5 minutes	3 (1.6%)
	5 to 15 minutes	89 (48.9%)
	15 to 30 minutes	20 (11.0%)
	More than 30 minutes	6 (3.3%)

Table III. Provider's practice and factors influencing uptake

		n (%)
Testing Offered	First trimester serum screen	106 (58.2%)
	Quad serum screen	62 (34.1%)
	Non-invasive prenatal screen (NIPS)	107 (58.8%)
	Amniocentesis	31 (17.0%)
	Chorionic Villus Sampling (CVS)	21 (11.5%)
	None	8 (4.4%)
	I do not recall what I was offered	24 (13.2%)
Points Addressed About Testing	What testing can detect	150 (86.2%)
	What testing cannot detect	78 (44.8%)
	Technology and methodology of how the tests are performed	83 (47.7%)
	Physical risks to the pregnancy	66 (37.9%)
	Beliefs and preferences about whether testing is right for you	50 (28.7%)
	Detection rates, false positives, false negatives	71 (40.8%)
	Interpretation of possible results	59 (33.9%)
	What to do after getting your results	47 (27.0%)
	Provider did not talk about the test	11 (6.3%)
Uptake of Testing*	First trimester serum screen	77 (72.6%)
	Quad serum screen	42 (67.7%)
	Non-invasive prenatal screen (NIPS)	84 (78.5%)
	Amniocentesis	8 (25.8%)
	Chorionic Villus Sampling (CVS)	2 (9.5%)
	I had genetic testing, but I'm not sure what type	20 (11.5%)
	I am not sure if I had genetic testing during my pregnancy	6 (3.4%)
	I did not have any testing done	25 (14.4%)
Factors Influencing Uptake*	Cost of testing	45 (25.9%)
	Desire to learn more about the health of the baby	118 (67.8%)
	Desire to learn the sex of the baby	36 (20.7%)
	Beliefs regarding genetic testing	28 (16.1%)
	Risks of testing	42 (24.1%)
	Because my healthcare provider recommended it	56 (32.2%)
	Because I had it done during my last pregnancy	26 (14.9%)
	Pressure from family/friends/community	5 (2.9%)

* percentages reflect only those who were offered testing

Table IV. Participant quiz responses

Survey Question	First Trimester Serum Screen	Quad Serum Screen	NIPS	US	Amnio	CVS	None	I Don't Know
Which test(s) will identify most pregnancies with Down Syndrome or similar chromosomal abnormalities?	63* (34.6%)	48* (26.4%)	90* (49.5%)	38 (20.9%)	82* (45.1%)	52* (28.6%)	0 (0.0%)	53 (29.1%)
Which test(s) is capable of diagnosing a pregnancy with Down syndrome or similar chromosomal abnormalities with 100% certainty?	11 (6.0%)	11 (6.0%)	18 (9.9%)	9 (5.0%)	63* (34.6%)	34* (18.7%)	43 (23.6%)	59 (32.4%)
Which test(s) will tell you if the pregnancy is a boy or a girl?	32 (17.6%)	23 (12.6%)	79* (43.4%)	132* (72.5%)	45* (24.7%)	29* (15.9%)	1 (0.6%)	24 (13.2%)
If you suspect your pregnancy has an increased risk of recessive conditions, such as sickle cell anemia or cystic fibrosis, which test(s) would provide you with more information?	17 (9.3%)	14 (7.7%)	35 (19.2%)	9 (5.0%)	49* (26.9%)	40* (22.0%)	4 (2.2%)	107 (58.8%)
Which test(s) will identify a pregnancy with a neural tube defect, like spina bifida?	23 (12.6%)	28* (15.4%)	28 (15.4%)	73* (40.1%)	46* (25.3%)	28 (15.4%)	2 (1.1%)	83 (45.6%)
Which test(s) pose a risk to the pregnancy and can result in a miscarriage?	1 (0.6%)	5 (2.8%)	2 (1.1%)	4 (2.2%)	128* (70.3%)	70* (38.5%)	3 (1.7%)	44 (24.2%)
Which test(s) will tell you for sure if your baby will be born healthy?	9 (5.0%)	6 (3.3%)	12 (6.6%)	14 (7.7%)	16 (8.8%)	11 (6.0%)	117* (64.3%)	41 (22.5%)
Which test(s) require another test to confirm the results if they come back with abnormal results?	79* (43.4%)	67* (36.8%)	68* (37.4%)	59* (32.4%)	21 (11.5%)	21 (11.5%)	1 (0.6%)	80 (44.0%)

* indicates correct answer

Table V. Participant knowledge scores between different variables

		Knowledge score (average ± SD)	p-value
Advanced Maternal Age Status	<35 years	29.0 ± 21.3	<0.001*
	≥35 years	41.2 ± 20.9	
Number of Children	0	28.6 ± 21.7	0.006*
	1+	37.5 ± 21.3	
Previously Met with a Prenatal Genetic Counselor	Yes	50.6 ± 21.3	<0.001*
	No	28.8 ± 21.2	
Provider Type	Genetics Provider	45.3 ± 22.8	0.008*
	Non- Genetics Provider	31.3 ± 21.4	
Time Spent Educating	0-5 mins	28.3 ± 19.4	0.056
	5 to 15 minutes	34.2 ± 22.4	
	15 or more minutes	39.5 ± 24.5	
Aneuploidy Testing Offered	None	18.0 ± 14.9	0.011*
	Screening	34.8 ± 20.9	
	Diagnostic Testing	31.9 ± 8.20	
	Both	44.3 ± 22.4	
Eligibility Criteria	Third Trimester	32.5 ± 21.3	0.919
	Gave Birth in Past 12 Months	32.9 ± 22.2	

* statistically significant at $p < 0.05$.

Table VI. Participants' responses to open-ended question

		n (%)	Examples
Provider experience	Positive experience	6 (14.3%)	"genetic counselors were very helpful"
	Negative experience	16 (38.1%)	"I wish I was given more information on the topic from the doctor rather than having to read about it"
	Incomplete patient education	8 (19.0%)	"I wish things had been explained more clear regarding dangers and what the tests actually entailed"
	No informed consent	4 (9.5%)	"the test was being performed without asking me"
	Directive provider	3 (7.1%)	"they were pushing for amnio"
	Lack of provider knowledge	3 (7.1%)	"the midwife I spoke to was very poorly informed"
Factors influencing testing	Insurance coverage/ cost	12 (28.6%)	"wish insurance provided coverage for more testing"
	Testing limitations	9 (21.4%)	"I had lost a twin, so that prevented me from receiving accurate results"
Participant characteristics	Information seeking	11 (26.2%)	"I wanted to be as prepared as possible"
	Deliberating testing options	6 (14.3%)	"I was very glad to have pursued amniocentesis with this pregnancy, though the decision was not easy- due to the risk, however small, of miscarriage...The NIPT test is great but there is still that fear of a false negative"
Testing experience	Positive experience	4 (9.5%)	"I was very pleased this was an option for me"
	Negative experience	4 (9.5%)	"after getting tested, my son was still born with a chromosomal defect"
	Beliefs that testing was not necessary	4 (9.5%)	"I declined all optional tests as I did not see the relevance to my personal situation"