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Making the most of the first prenatal visit: The challenge of expanding prenatal genetic testing options and limited clinical encounter time

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Abstract

Objective: Advances in prenatal genetics place additional challenges as patients must receive information about a growing array of screening and testing options. This raises concerns about how to achieve a shared decision-making process that prepares patients to make an informed decision about their choices about prenatal genetic screening and testing options, calling for a reconsideration of how healthcare providers approach the first prenatal visit.

Methods: We conducted interviews with 40 pregnant women to identify components of decision-making regarding prenatal genetic screens and tests at this visit. Analysis was approached using grounded theory.

Results: Participants brought distinct notions of risk to the visit, including skewed perceptions of baseline risk for a fetal genetic condition and the implications of screening and testing. Participants were very concerned about financial considerations associated with these options, ranking out-of-pocket costs on par with medical considerations. Participants noted diverging priorities at the first visit from those of their healthcare provider, leading to barriers to shared decision-making regarding screening and testing during this visit.

Conclusion: Research is needed to determine how to restructure the initiation of prenatal care in a way that best positions patients to make informed decisions about prenatal genetic screens and tests.

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1 | INTRODUCTION

Rapid advances in cell-free DNA (cfDNA) screening have significantly affected the delivery of prenatal care. This prenatal genetic screen provides more accurate information about the risk of Trisomy 21, Trisomy 18 and Trisomy 13 compared to other maternal serum marker screens. In addition, this test can also analyze sex chromosomes and screen for microdeletions and rare aneuploidies.[1] New clinical practice guidelines recommend that, rather than just the subset of women considered to be at increased risk due to age or reproductive history, all pregnant patients should be offered prenatal genetic screening or diagnostic testing.[2] With such changes in the delivery of prenatal care come new benchmarks for healthcare providers to ensure patients have the information and resources necessary to make informed, values-based decisions about their prenatal genetic screening and testing options.

An optimal time to discuss prenatal genetic screening and testing options is the first prenatal visit. The volume and nature of information about identifying fetal genetic risks present unique challenges for the initial visit. This is due to the varying health literacy levels and numeracy skills that patients bring to the visit, as well as the personal and value-laden nature of prenatal genetic-testing decisions.[3–6,7] Other information necessary to convey at this first prenatal visit may include management of preexisting medical conditions, medication use, vaccination, and other emerging concerns for the pregnancy (eg, Zika virus, COVID-19).[8] As a result, the first prenatal visit is unique in that a large volume of information must be effectively conveyed and discussed between the patient and her provider.

This situation raises the question of how to best structure effective, patient-centered, and evidence-based conversations about prenatal screens and tests during this initial visit. Shared decision-making is an optimal way to structure complex healthcare discussions.[9–11] Shared decision-making involves a collaborative agenda setting process in which the priorities of the patient and the provider are shared to structure the visit discussions.[12] Despite the recognized potential to provide key support for prenatal genetic screening and testing options, little is known about how to apply this approach in the prenatal care setting, particularly, at the first prenatal visit.

To help patients make informed decisions about prenatal genetic screens and testing, it is critical that procedures are in place to ensure that patients have the education and resources necessary. Improving patient experience, healthcare communication, and evidence-based delivery are major ways to increase healthcare quality and safety in the United States. Therefore, this study was conducted to gain a greater understanding of the factors that impact shared decision-making and discussions about prenatal genetic screening and testing during the initial visit.

2 | METHODS

Interviews were conducted to explore patients' decision-making regarding the options of prenatal genetic screening and diagnostic testing. Study procedures took place at outpatient prenatal care clinics within a healthcare system that offered cfDNA primarily to the

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high-risk obstetric population (based on reproductive history and/or advanced maternal age; AMA) and as defined by contemporaneous clinical practice guidelines. All research procedures were approved by the [deidentified] Institutional Review Board.

Participants were sampled from a larger, randomized control trial examining the impact of a point-of-care shared decision-making tool compared to standard care on patients' decisions about prenatal genetic screening and diagnostic testing (the Non-Invasive prenatal genetic testing Education Support Tool [NEST] study).[13] We utilized a purposeful recruitment approach by taking a random sample of NEST study participants seeking equal representation from those who were part of the intervention arm and control arm of the study. At the time of recruitment for this study, the NEST study was and continues to be recruiting. Recruitment efforts were made to match the population of the larger NEST study, of which, at the time, 89.1% self-identified as Caucasian and 81% as having college- or graduate-level education (higher education). Inclusion criteria included women who were enrolled in and completed data collection as part of the NEST study, were 18 years of age or older, were in the third trimester of pregnancy or had delivered in the past 12 months, had received outpatient prenatal care at the [deidentified] Health System, were English speaking, and could provide consent for research participation. Eligible participants were sent a recruitment letter through their previously approved method of communication for the NEST study.

Telephone interviews were conducted by two research team members with experience in research among this patient population. Data collection was done using an interview guide developed by experts in obstetrics, prenatal genetics, ethics, and medical decision-making and based upon the authors' prior work. The guide contained open-ended questions regarding informational needs and preferences in the decision-making process at the onset of prenatal care and how those factors may have changed over time during the course of the pregnancy and/or with decisions about prenatal genetic screening and diagnostic testing. Individuals who were randomized to the intervention arm of the NEST study were asked an additional set of questions about the role of the intervention in their decision-making (those data reported separately).

Data analysis was conducted by an iterative and process of progressive data immersion, open coding, documentation, and theme identification consistent with grounded theory.[14,15] Coding was performed independently by two coders. The coders met at regular intervals to review the code book with any emergent themes, identify and resolve any coding discrepancies, and determine the point where thematic saturation was achieved. NVivo12 was used in conjunction with memos to record emergent themes and insights into data interpretation. Interviews from the intervention and control arm were analyzed separately; then, themes were compared across groups during and at the conclusion of the analysis.

3 | RESULTS

A total of 77 eligible participants were approached for study participation. Participants were enrolled sequentially. Thematic saturation was achieved with 40 interviews, with 20 participants from the intervention arm and 20 participants from the control arm. The

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mean age of respondents was 32 ± 4 years (Table 1). The majority had a prior pregnancy (55%, *n* = 22), self-reported as Caucasian (90%, *n* = 36), and had a college or higher educational degree (75%, *n* = 30). Nineteen had undergone cfDNA screening during the current pregnancy (47.5%, *n* = 19). All participants who had undergone cfDNA screening received counseling from a maternal-fetal specialist or genetic counselor as is the practice of [deidentified] health system.

Qualitative analysis identified four major themes relating to pregnant patients' needs, priorities, and baseline perceptions regarding prenatal genetic screening and diagnostic testing at the onset of their prenatal care (see Table 2 for supplemental data regarding each theme). No difference in themes was noted among the intervention and control groups.

3.1 | Perceptions of risk for a fetal genetic condition

Participants brought specific beliefs to the first prenatal visit about their risk of having a pregnancy affected with a genetic condition. These beliefs served as an anchoring point to consider the utility of prenatal genetic screening or testing. Beliefs around maternal age were found to be a significant factor. The majority of participants referenced 35 years of age as a primary threshold for determining the chance of the pregnancy being affected by fetal genetic condition. This belief was seen as the starting point for a series of other considerations, including, the perceived benefit of genetic screening or testing, and which approach(s) the patient might take to obtain that information. As described by this participant, "*Going into the second child, since I will be over 35, just because of my age alone, I would just assume the doctor would recommend more testing than the first child. And I, myself, just kind of guessed there would probably be a need to do more tests then the first child just because of that [age]*" (10-23). Baseline beliefs about maternal age extended to the probability of any congenital abnormality, including those not linked with a genetic variant. "*The realization that I am a higher maternal age and that puts me at a great risk of having a child with some kind of abnormality*" (10-05).

These framing beliefs about maternal age and risk influenced how women approached the first prenatal visit. Some, because they were younger than 35 years of age, did not consider the possibility of genetic screening or testing before the first visit, including the potential benefits of screening or testing (eg, preparation for the birth of a child with a genetic condition) or values-based factors that go into such decisions. For instance, one participant, who did not consider the possibility of screening or testing prior to her first prenatal visit, felt under prepared when learning of her options during the appointment: "*I would say just I think my initial visit was just kind of um thinking that a lot of the things didn't affect me or weren't gonna be an issue as I wasn't like high risk and my age wasn't a factor*" (30-08). This belief about maternal age had a further impact on the patients' perception of their ability to make the choice to accept or decline screening or testing. As described by this participant, who felt that she did not have a choice in undergoing screening or testing, "*I knew it was required or highly recommended if you were over 35 and optional if you were younger and that's pretty much it*" (120-03).

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3.2 | Considering risks of prenatal genetic screening and testing

Participants also brought specific beliefs about the risks involved with screening and testing to the first prenatal visit. Perception of risk to the fetus as a result of diagnostic testing was a lead factor in these considerations. As described by these participants, "*I think the risks for me were the most important…*" (10-15) and "*I just wanted to make sure that whatever we did in terms of testing was safe for me and the baby, so that's kind of how we made the decision for what we would have done*" (10-17). Participants placed some types of screening and testing as opposite ends of a risk spectrum. While tests such as cfDNA screening were considered "safe," diagnostic testing by means of amniocentesis or chorionic villus sampling was considered as "risky," "dangerous," and "invasive." As described by this participant, "*I've heard it's [amniocentesis] pretty dangerous and usually the risk is not really worth doing it is what I hear, unless you get a positive result from a first test like a blood test.*" (10-08). In fact, the perceived "safety" of screening compared to diagnostic tests was a compelling factor in why some participants elected for screening. As described by this participant, "*It wasn't invasive. It was only gonna test for these things and you know it wouldn't cause any harm to the baby and so I thought, ok, knowledge is power […]. So knowing that it wasn't going to cause any harm, I was like, 'Yeah, let's do it'*" (50-09).

3.3 | Financial risks as a component of decision-making

Several participants commented that any out-of-pocket costs would be a major determining factor in the decision to consider and then undergo genetic screening or testing. For this reason, they felt it was essential for healthcare providers to discuss insurance coverage and out-of-pocket expenses at the same time as discussing the medical considerations and options of screening and testing. As described by this participant, the costs associated with screening or testing would require significant compromises for her family, "*I think cost is one of the biggest issues that comes with any health care and people have to decide do I … how badly do I need this test versus do I need groceries this month?*" (50-10). Another participant reflected on the impact of existing healthcare costs for her and her family: "*I just didn't want a surprise bill. There's just so many dollar signs enough as it is*" (10-11). For this participant, information about the out-of-pocket costs for screening was just as important as information about the screen's characteristics (eg, what conditions were screened for, detection rate). As she said:

> *I think also something that would have been good to know ahead of time would have been how much it costs. To have that information, […] it might sway people's decision one way or another. You hope to not have to make decisions like that because of money but […] I think knowing that it's not covered by insurance in advance […] So, if they know that financially, it's just not going to be for them, then they can maybe not read about it and get excited about it as something they want to do and not like it's, "Just, oh shoot. It's a $500 test." I do not have that in my life laying around* (120-06).

Several participants noted that financial considerations were so important to their choice about genetic screening and testing that this topic should have been addressed prior to the first prenatal visit. As one of the participants continued stated, "*When patients, in general, hear they're having a child, they're excited and, then also freaked out because of the cost …*

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People should know their insurance plans, but they don't until they have a $5000 bill and realize that's their deductible. So, I think if they had a list of all the tests on a sheet of paper that's recommended along with the codes […] so then they can decide, ok, this test is a test I feel is really necessary. […]." She also raised the idea of the healthcare provider taking the initiative to provide this information: "*The doctor could go ahead and have one of their staff check to make sure it's in network and get approval ahead of time. That would be easier. So, when they go into the appointment, they say, 'Alright. We already have approval from your insurance. They will cover this 100%'*" (50-10).

3.4 | Priorities at the first visit and challenges to developing a collaborative agenda

Many patients felt a mismatch between their priorities at the first prenatal visit and those of their providers. For instance, many participants presented to the visit wanting reassurance of a viable intrauterine pregnancy. This was particularly relevant for women who were experiencing pain or bleeding in the first weeks of pregnancy or had a prior history of a complicated pregnancy or a preexisting medical condition. For this participant, previous medical concerns outweighed the topic of prenatal screening or testing at the first prenatal visit: "*I have [specific medical condition] so I was wondering if the pregnancy was going to last. So, I was like, well let's wait till that happens. Let's wait till I'm farther in to make a decision [about screening and testing] and things like that*" (70-13).

Because many women felt that the purpose of the first visit was to see or hear the fetal heart beat, they felt unprepared to discuss other prenatal care topics that their provider felt was important to address that visit. As described by this participant, "*I think because your first visit is at week 10. You know, there's a lot you wanna cover… you know when you're having questions at the beginning and you wanna hear the heart beat for the first time. So, it might not seem like that [prenatal genetic screening and testing] is at the top of your priority list to discuss.*" (100-01). As a result, many felt there was a mismatch between their informational priorities and what they felt their healthcare provider wanted to discuss during the visit. Some participants felt their provider did not spend the expected amount of time discussing their needs. As this participant said, "*I guess with my experience, my husband and I both felt rushed in that first appointment. We didn't feel like all of our questions were answered*" (100-02). Like many other participants she felt underprepared for that visit: "*But we also may not have had the right questions to ask at the first appointment, either…*" (100-02). In addition, like other patients, she did not feel prepared for the nature or volume of information conveyed about fetal genetic risk and assessment at that initial visit: "*That first visit is such a whirlwind and it's so much information to take in and you may not remember everything you want to remember.*" (70-05) Similarly, another participant stated that having reliable information before the first visit would have made her feel better prepared for any screening and testing decisions that needed to be made at the first visit:

> "*If there was like a […] reliable source rather than Googling stuff about the different prenatal test and things like that so they could get good information going in [to the visit] and, you know, kind of already have maybe thought about some things before they get asked this question so that they are not just put on the spot like, 'Do you want to do this or not?' So they don't feel overwhelmed with thinking it's a decision just based on a 5-10 minute visit with the doctor*" (30-09).

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4 | DISCUSSION

Agenda setting is an integral part of the shared decision-making process and is particularly important at the first prenatal visit, as the discussions during this visit can lay the framework for prenatal testing which may shape the course and outcomes of the pregnancy.[6,16,17] Our study highlights the need to revisit how prenatal care is established, with specific emphasis on the first prenatal encounter, which starts the healthcare decision-making process for the rest of the pregnancy. Many participants felt unprepared to engage in discussions about prenatal genetic screening and diagnostic testing at this visit. Yet, this is also the visit in which healthcare providers seek information to identify if a patient may be at increased risk of a genetic condition or obstetrical complication and, in response, outline recommended screens and tests to assess that risk.[18] Our findings suggest that patients and providers have different priorities at the first prenatal visit and that these differences may interfere with shared decision-making.

Another potential barrier to effective agenda-setting was the patient's general knowledge as well as their perceived risks with respect to prenatal genetic screening and testing. Prior studies have shed light on knowledge and health literacy levels of patients who present for prenatal care, demonstrating the struggles faced by patients to understand concepts of risk associated with prenatal genetic screening.[5,6] Our study found that participants had outdated notions of risk centered on the notion of advanced maternal age as a prime determinant of genetic risk. This stands in contrast to the prevailing stance to do away with maternal age as an indication for screening or testing.[19]

Our findings also showed that participants have inaccurate perceptions of risk associated with diagnostic testing and the chance of pregnancy loss associated with amniocentesis and chorionic villus sampling. The risk of a pregnancy loss or fetal injury is viewed by healthcare providers as low, often cited as ≤1% when performed by a trained practitioner.[20,21] Yet, participants had very different notions at the onset of their prenatal care, describing these diagnostic testing procedures as "dangerous," "risky," and "not safe." Their opinions were based largely on information from social media, the Internet, and shared experiences of friends and relatives before the first visit. These findings indicate that investment of time and effort is required at the initial prenatal visit to determine patients' knowledge levels and how these influence how patients will approach the topic of prenatal screening. This process may be facilitated by pre-visit questionnaires and decision aids that clarify patient and provider expectations of the nature and detail of topics that will be addressed at the visit, followed by targeted and individualized educational materials for review prior to the visit. There may also be a role for telehealth pre-visit appointments in which a healthcare team member begins the educational process in preparation for the first visit.

In addition, we found that out-of-pocket costs associated with screening or testing comprised a significant consideration that shaped the decision to obtain genetic information about the fetus and what modality to utilize. While some prior studies have evaluated patients' financial considerations associated with prenatal genetic testing, most have focused on cost-effective analysis or on the perspectives of clinicians.[22–29] Therefore, further research is

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needed to understand how women of different demographic and socioeconomic groups may view the financial aspects of prenatal genetic screening, and the impact of those views on decisions to use or decline new screens or tests.

Our study provides insight into the challenges associated with the exchange of an increasing volume of complex health information at the initiation of prenatal care. Limitations of our study are that findings were based on the perspectives of a small cohort of women who presented for prenatal care in the United States. Most of these women were in their 30s, had undergone a prior pregnancy, had self-reported as Caucasian, and had a college or higher education level. In addition, we enrolled women who were in their the third trimester of pregnancy or postpartum, a factor that may have introduced a recall bias. Moreover, these women were also part of a larger study which focused on decision-making; thus, they may have been uniquely attuned to issues of communication and decision-making. Consequently, further research is needed to determine whether the experiences of the women in this study correlate with those of women who represent different patient populations with respect to age, race, reproductive history, and access to healthcare.

5 | CONCLUSIONS

Patient-centered and evidence-based approaches to the first prenatal visit will grow in importance as new prenatal genetic screening and diagnostic testing options increase the volume of information that needs to be conveyed to patients. Further research is needed to determine how to present and discuss information at the first prenatal visit in a way that best supports the shared decision-making process in a way which meets the priorities of both patients and healthcare providers.

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CONFLICT OF INTEREST

Dr. Rose discloses the following relationship with industry that are not directly relevant to the topic: SR received speaking honorariums and travel funding within the past three years from Siemens Healthineers, Panagora Pharma, Healthcare Information and Management Systems Society, Inc. (HIMSS), Next Generation Patient Experience (NGPX), and healthcare systems in Sweden and Saudi Arabia on topics related to public health, bioethics and health policy. She served on the Ethics Committee for the American Society of Clinical Oncology (ASCO), a non-profit.

The authors declare no potential conflict of interest.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available on request from the corresponding author. The data are not publicly available due to privacy or ethical restrictions.

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REFERENCES

1. Gil MM, Quezada MS, Bregant B, Ferraro M, Nicolaides KH. Implementation of maternal blood cell free DNA testing in early screening for aneuploidies. Ultrasound Obstet Gynecol. 2013;42(1):34–40. 10.1002/uog.12504. [PubMed: 23744609]
2. Gregg AR, Skotko BG, Benkendorf JL, et al. Noninvasive prenatal screening for fetal aneuploidy, 2016 update: a position statement of the American college of medical genetics and genomics. Genet Med. 2016;10:1056–1065. 10.1038/gim.2016.97.
3. Shieh C, Halstead JA. Understanding the impact of health literacy on Women's health. J Obstst Gynecol Neonatal Nurs. 2009;38(5):601–610. 10.1111/j.1552-6909.2009.01059.x.
4. Seven M, Akyüz A, Eroglu K, Daack-Hirsch S, Skirton H. Women's knowledge and use of prenatal screening tests. J Clin Nurs. 2017;26(13–14):1869–1877. 10.1111/jocn.13494. [PubMed: 27487389]
5. Sheinis M, Bensomon K, Selk A. Patients' knowledge of prenatal screening for trisomy 21. J Genet Couns. 2018;27(1):95–103. 10.1007/s10897-017-0126-3. [PubMed: 28707139]
6. Shea TL. Informed decision making regarding prenatal aneuploidy screening. J Obstet Gunecol Neonatal Nurs. 2020;49(1):41–54. 10.1016/j.jogn.2019.11.001.
7. Agatisa PK, Mercer MB, Mitchum A, Coleridge MB, Farrell RM. Patient-centered obstetric care in the age of cell-free fetal DNA prenatal screening. J Patient Exp. 2018;5(1):26–33. 10.1177/2374373517720482. [PubMed: 29582008]
8. American Academy of Pediatrics, American College of Obstetricians and Gynecologists. Guidelines for Perinatal Care. 8th Ed. Chapter 6: antepartum care. Washington, DC: The American College of Obstetricians and Gynecologists (150–226). https://reader.aappublications.org/guidelines-for-perinatal-care-8th-edition/168. Accessed March 4, 2020.
9. Howard H, Freeman K, Clark K. U.S. survey of shared decision making use for treating pregnant women presenting with opioid use disorder. Subst Use Misuse. 2019;54(13):2241–2250. 10.1080/10826084.2019.1644524. [PubMed: 31349761]
10. Wu YL, Lu HC, Chen KJ, Chang SI. Application of shared decision making on birth choice in a primigravida woman with congenital adrenal hyperplasia. Hu Li Za Zhi. 2019;66(5):8–85. 10.6224/JN.201910_66(5).11.
11. Popreczny AJ, Stocking K, Showell M, Duffy JMN. Patient decision aids to facilitate shared decision making in obstetrics and gynecology: a systematic review and meta-analysis. Obstet Gynecol. 2020;135(2):444–451. 10.1097/AOG.0000000000003664. [PubMed: 31923056]
12. Abbasgholizadeh-Rahimi S, Archambault PM, Ravitsky V, et al. An analytical mobile app for shared decision making about prenatal screening: protocol for a mixed methods study. JMIR Res Protoc. 2019;8(10):e13321. 10.2196/13321. [PubMed: 31596249]
13. NEST Study ensuring patients' informed access to noninvasive prenatal testing (NEST) (ClinicalTrials.gov Identifier: NCT03420274) https://clinicaltrials.gov/ct2/show/NCT03420274?term=NEST&draw=2&rank=4.
14. Corbin J, Strauss A. Basics of Qualitative Research: Techniques and Procedures for Developing Grounded Theory. (3rd)Thousand Oaks, CA: Sage Publications Inc; 2008. doi:10.4135/9781452230153
15. Ryan GW, Bernard R. Techniques to identify themes. Field Methods. 2003;15(1):85–109. doi:10.1177/1525822X02239569.
16. Farrell RM, Agatisa PK, Michie MM, Greene A, Ford PJ. The personal utility of cfDNA screening: pregnant patient experiences with cfDNA screening and views on expanded cfDNA panels. J Genet Couns. 2020;29(1):88–96. 10.1002/jgc4.1183. [PubMed: 31680382]
17. Berhardt BA, Geller G, Doksum T, et al. Prenatal genetic testing: content of discussions between obstetric providers and pregnant women. Obstet Gynecol. 1998;91(5 Pt1):648–655. [PubMed: 9572205]
18. Lee S, Holden D, Webb R, Ayers S. Pregnancy related risk perception in pregnant women, midwives & doctors: a cross-sectional survey. BMC Pregnancy Childbirth. 2019;19(1):335. 10.1186/s12884-019-2467-4. [PubMed: 31558157]

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19. American College of Obstetrics and Gynecology. Practice bulletin no. 163: screening for fetal aneuploidy. Obstet Gynecol. 2016;127(5):e123–e137. 10.1097/AOG.0000000000001406. [PubMed: 26938574]
20. Monni G, Pagani G, Illescas T, Stagnati V, Iuculano A, Ibba RM. Training for trasnabdominal villous sampling is feasible and safe. Am J Obstet Gynec. 2015;213(2):248–250. 10.1016/j.ajog.2015.04.019. [PubMed: 25912301]
21. Beta J, Zhang W, Geris S, Kostiv V, Akolekar R. Procedure-related risk of miscarriage following chorionic villus sampling and amniocentesis. Ultrasound Obstet Gynecol. 2019;54(4):452–457. 10.1002/uog.20293. [PubMed: 30977213]
22. Abdo N, Ibraheem N, Obeidat N, et al. Knowledge, attitudes, and practice of women toward prenatal genetic testing. Epigenet Insights. 2018;11. 10.1177/2516865718813122
23. Suskin E, Hercher L, Aaron KE, Bajaj K. The integration of noninvasive prenatal screening into the existing prenatal paradigm: a survey of current genetic counseling practice. J Genet Couns. 2016;25(5):1032–1043. 10.1007/s10897-016-9934-0. [PubMed: 26879922]
24. Kuppermann M, Norton ME, Thao K, et al. Preferences regarding contemporary prenatal genetic tests among women desiring testing: implications for optimal testing strategies. Prenat Diagn. 2016;36(5):469–475. 10.1002/pd.4808. [PubMed: 26946227]
25. Kaimal AJ, Norton ME, Kuppermann M. Prenatal testing in the genomic age: clinical outcomes, quality of life, and costs. Obstet Gynecol. 2015;126(4):737–746. [PubMed: 26348190]
26. Nshimyumukiza L, Menon S, Hina H, Rousseau F Reinharz D. Cell-free DNA noninvasive prenatal screening for aneuploidy versus conventional screening: a systematic review of economic evaluations. Clin Genet. 2018 94(1):3–21. doi: 10.1111/cge.13155. Epub 2018 Jan 25 [PubMed: 29030960]
27. Benn P, Curnow KJ, Chapman S, Michalopoulos SN, Hornberger J, Rabinowitz M. An economic analysis of cell-free DNA non-invasive prenatal testing in the US general pregnancy population. PloS One. 2015;10(7):1–12. 10.1371/journal.pone.0132313.
28. Fairbrother G, Burigo J, Sharon T, et al. Prenatal screening for fetal aneuploidies with cell-free DNA in the general pregnancy population: a cost-effectiveness analysis. J Matern Fetal Neonatal Med. 2015;7058:1–5. 10.3109/14767058.2015.1038703.
29. Walker BS, Nelson RE, Jackson BR, Grenache DG, Ashwood R, Schmidt RL. A cost-effectiveness analysis of first trimester non-invasive prenatal screening for fetal trisomies in the United States. PloS One. 2015;10(7):1–20. 10.1371/journal.pone.0131402.

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What is already known about this topic?

- Pregnant patients should be prepared to make informed, values-reflective decisions about their prenatal genetic screening and diagnostic testing options.
- These are often complex decisions as a function of patients' health literacy, baseline knowledge and perceptions of genetic conditions and risk, and patient values.
- Advances in molecular sequencing has increased the volume of information that can be conveyed by prenatal genetic screens and diagnostic tests, increasing the complexity of such decisions for patients and healthcare providers positioned to support a shared decision-making process about their use.

What does this study add?

- Despite a change in how healthcare providers view the risk of a fetal genetic condition affecting a pregnancy and recommendations to offer aneuploidy screening and diagnostic testing to all patients, patients maintain outmoded and skewed notions of advanced maternal age and procedure-related risks as primary drivers in their decision-making about screens and diagnostic tests.
- Financial considerations, specifically patients' out of pocket costs, are also a significant driver in prenatal genetic screening and diagnostic decisions and a lead priority for patients in initial discussions about whether and how to obtain genetic information about the fetus.
- The status of the pregnancy (eg, seeing or hearing evidence of an intrauterine pregnancy with cardiac activity) is a primary concern for patients at the first prenatal visit, often taking precedence over the priorities of healthcare providers in addressing the medical considerations of genetic screening and diagnostic testing during this initial clinical encounter.

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TABLE 1

Demographics

Demographic factor	***N* (%)**
AMA	11 (27.5%)
Non-AMA	29 (72.5%)
No prior pregnancy	18 (45%)
Prior PREGNANCY	22 (55%)
Race	
White	36 (90%)
Black	3 (7.5%)
Asian	1 (2.5%)
Other	1 (2.5%)
Education level	
High school or GED	2 (5%)
Associate, technical degree	8 (20%)
College degree	14 (35%)
Graduate or professional degree	16 (40%)
Received cfDNA testing	19 (47.5%)
No cfDNA testing	21 (52.5%)

Note: Percentages in the table do not equal 100% due to a ‘choose all that apply format’ of the question and one multi-rational patient.

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TABLE 2

Supplemental data

Theme	Illustrative quote
Perceptions of risk for a fetal genetic condition	"I felt like it was something I really did not need. So I wasn't looking into it." (100-03) "Fortunately, my partner and I were not genetic predisposed to what they were looking for either. So, we felt like ok, given our family history…" (50-09) "So my husband and I had decided that we did not want to go through with a lot of the testing we had available to us in terms of the blood work because were both healthy …" (120-06) "I knew it was required or highly recommended if you were over 35 and optional if you were younger and that's pretty much it." (120-03)
Considering risks of prenatal genetic screening and testing	"I just wanted something that was non-invasive that was not gonna be a health risk to me or the baby." (30-10) "The NIPT testing was the big one- I think it was the amniocentesis was the other one and that was more like I really did not want to do it unless I absolutely had to just given the higher chances of risk involved with doing something like that versus the NIPT testing just drawing the blood form the mom's arm and like the amniocentesis is more heavily involved for the child." (120-06) "So I really had not thought about much about any kind of genetic testing except that (amnio) and I knew I would reject that (amnio) since it was invasive." (10-15) "I think the risks for me were the most important like if they were an invasive procedure or not." (10-15)
Financial risks as a component of decision-making	"It wasn't gonna make a difference [for the pregnancy outcome. […] The cost of the testing alone is very expensive. […] So that kind of sealed my decision." (30-09) "But I had to pay out of pocket for it then and so now and because I'm older, it was covered by my insurance, so it was like free to me. So why not just do it?" (30-11) "Not knowing exactly what the cost of the testing was going to be … We knew we were probably going to do it. We had made the decision going in that we were going to do it regardless, but that was one of the things that was kind of like, well, we are not exactly sure how much even before insurance paid in. Just not knowing we could be on the hook for this amount to this amount depending on this or that. That information wasn't available." (120-03) "One of the factors was like if insurance would cover it. That was a big factor of us getting it 'cause I knew it was expensive." (120-02) "It was a couple of things. One was insurance coverage just because we were not terribly worried about it but it was nice to have the peace of mind about it, the insurance coverage. And then the risk to the baby. I'd say were like the two main things." (10-24)
Priorities at the first visit and challenges to developing a collaborative agenda	"This is my second pregnancy. The first pregnancy I believe they kind of went over everything but … being first time pregnancy and mom, a lot of it seemed kind of foreign." (30-08) "I mean, when you first find out your pregnant, I do not think that that's a thing that people think about… a possibility of any genetic issues." (100-02) "When you are in the beginning of the pregnancy and you have to make all of those decisions and you are still all excited or maybe unsure, or like just getting use to this new part of your life, I think it can be like a lot of pressure to make all those decisions at once." (10-02) "I think just continuing to keep it simple. I really think a handout, like I said just really simple and you could go over it together and ask any questions at that point. And making sure the doctor reiterates that it's ok to ask questions 'cause some people are nervous at that first appointment. Kind of a nerve-racking appointment for sure." (120-02) "The first visit, I think you kinda just wanna know there's a heartbeat is the big thing." (10-11) "I was a bit surprised by the pregnancy even. Just because like the duration of my first pregnancy, like you know trying to have a baby and this one I was just a surprise … I was pregnant. […] I had a toddler so that was part of it. I do not know if I thought about too much in advance like with the first one [first pregnancy] […] cause I had a lot of other things going on." (10-11) "You know when you are pregnant your brain's not always working 100%." (10-08)

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