

HHS Public Access

Author manuscript *Obstet Gynecol.* Author manuscript; available in PMC 2017 June 01.

Published in final edited form as:

Obstet Gynecol. 2016 June ; 127(6): 1145–1152. doi:10.1097/AOG.00000000001433.

Patient–Health Care Provider Conversations About Prenatal Genetic Screening: Recommendation or Personal Choice

Laura C. Colicchia, $MD^{1,2}$, Cynthia L. Holland, MPH^2 , Jill A. Tarr, MSW^2 , Doris M. Rubio, PhD^3 , Scott D Rothenberger, PhD^3 , and Judy C. Chang, MD, $MPH^{1,2,4}$

¹University of Pittsburgh Medical Center

²Magee-Womens Research Institute

³University of Pittsburgh Center for Research in Health Care Data Center

⁴Departments of Obstetrics, Gynecology, and Reproductive Science and Internal Medicine and Center for Research in Health Care, University of Pittsburgh School of Medicine

Abstract

Objective—To assess how obstetric health care providers counsel patients regarding prenatal genetic screening, and how these conversations influence patients' screening decisions.

Methods—This cohort study analyzed transcripts and audio recordings of 210 first prenatal visits, collected as part of a larger study on patient-provider communication. Conversations were coded in an iterative process to determine compliance with American College of Obstetricians and Gynecologists (the College) prenatal genetic screening recommendations and to identify recurrent themes. Chi-squared, nonparametric tests, and logistic regression were used to determine the effects of discussion elements on screening decisions. Qualitative analysis was performed for genetic screening content.

Results—The study included 210 patients and 45 health care providers. Health care providers offered genetic screening at 90% of visits; 78% of women chose genetic screening. Few conversations (1.5%) included all College-recommended topics. Inclusion of College-recommended topics did not affect women's screening choices. Conversations about screening for fetal aneuploidy lasted 1.5 minutes on average (range 0.12 to 7.05 minutes). Recurrent themes identified included clarifying that screening results are not diagnostic (51% of conversations), emphasizing that screening results to guide decisions about diagnostic testing or termination (37% of conversations). Health care providers described screening results as "high or low risk" in 67% of conversations discussing risk and quantitatively (ie, 1 in 100 or 1%) in 33%.

Conclusions—Although the majority of patients were offered and underwent screening, most health care providers' counseling did not adhere to College recommendations.

Corresponding Author: Laura Colicchia, MD, 300 Halket St., Pittsburgh, PA 15213, 412-641-5466, ; Email: colicchialc@upmc.edu From the University of Pittsburgh Medical Center, Magee-Womens Hospital, Pittsburgh, PA.

Presented at The Society for Reproductive Investigation 62nd Annual Meeting, San Francisco, CA, March 26, 2015.

Introduction

Chromosomal anomalies, most commonly Down Syndrome, complicate 1 in every 600 births in the United States.¹ The American College of Obstetricians and Gynecologists (the College) recommends that all pregnant women should be offered fetal aneuploidy screening in the first or second trimester.² At present, the primary screening modalities for low risk patients include the first trimester screen and multiple marker screening.³ When discussing screening tests, College guidelines suggest that health care providers specifically address (1) detection and false-positive rates, (2) advantages, disadvantages and limitations of the screening tests, and (3) the option of diagnostic testing.² Counseling should be nondirective, with the goal of allowing the patient to make an informed choice to pursue noninvasive screening, invasive testing, or no testing.^{4,5}

While most obstetric health care providers report routinely offering prenatal genetic screening to all their patients, prior studies suggest that many patients miss the opportunity for screening, and many health care providers are not aware of details such as the false positive or negative rate of screening tests.^{6,7} Furthermore, many women who do undergo testing later report being unaware of what the test was for or what the results meant.^{8–10}

The purpose of this study was to use audio-recorded prenatal visits to assess how obstetric health care providers currently counsel patients about prenatal genetic screening, and how this conversation influences patients' screening decisions. We hypothesized that inclusion of more College-recommended counseling topics in screening discussions would increase the number of women choosing prenatal genetic screening.

Materials and Methods

This cohort study is part of a larger study of patient-provider communication in prenatal care.^{11–13} We analyzed audio recordings of first prenatal visits at three urban clinic sites in Pittsburgh, Pennsylvania from 2011 to 2014. Obstetric health care providers at the clinics include obstetrics and gynecology residents, obstetrics and gynecology faculty and midlevel health care providers. Each study site serves a racially diverse population of women, 50–100% of whom are on medical assistance. IRB approval was obtained from the University of Pittsburgh Institutional Review Board.

Health care provider and patient participants were approached for participation prior to office sessions. We limited the number of patients enrolled per health care provider to ten. Exclusion criteria for patient participants included non-English speaking women and women younger than 18 years of age. Exclusion criteria for this analysis included women presenting for their first prenatal visit after 20 weeks gestation (beyond the recommended timeframe for prenatal genetic screening) and women who reported undergoing genetic screening at an outside institution prior to presenting for care at our clinic. After obtaining informed consent from both health care providers and patients, initial prenatal visits were audio recorded in their entirety. Patients and health care providers were consented to participate in a patient-provider communication study and were aware of the recording; they were not made aware of any specific topical focus including the focus of this analysis on genetic screening

conversations. Demographic information was obtained from patients and health care providers prior to the visit. Chart review of the electronic medical record was performed to determine which, if any, genetic tests the patient had ultimately completed.

Our institutional protocol is to offer prenatal genetic screening to all patients at first prenatal visits. Genetic counseling is available for patients who have additional questions or desire diagnostic testing. A question in the first prenatal visit template in our electronic medical record prompts health care providers to document whether and what type of testing was offered. All health care providers are encouraged to follow ACOG Practice Bulletin 77: Screening for Fetal Chromosomal Anomalies for counseling guidelines, and this practice bulletin is reviewed with residents in the clinic on a rotating basis along with other bulletins. Attendings in the clinic precept patients but do not always directly observe resident or midlevel patient interactions.

Recordings were transcribed and reviewed to confirm accuracy. Transcripts were reviewed in their entirety for any discussion of genetic testing, including first trimester screening, multiple marker screening, amniocentesis or CVS, or cell-free fetal DNA screening. If genetic testing was discussed, we reviewed which tests were offered, the duration of the discussion, who initiated the discussion, if the patient decided to have genetic testing, and the inclusion of College-recommended discussion topics (detection rate, false positive rate, advantages, disadvantages and limitations of screening and the availability of diagnostic testing). We limited our analysis to screening for fetal chromosomal anomalies and did not assess transcripts for discussion of cystic fibrosis, tay sachs or other carrier testing.

We performed a qualitative analysis of genetic screening conversations to identify recurrent themes. All transcripts were coded in an iterative process by one of the authors (L.C.C.) until thematic saturation was achieved. The final codebook was developed and then applied to the remaining transcripts. A second author (J.C.C.) independently coded 40 randomly selected transcripts (20%), and inter-rater reliability was calculated using Cohen's kappa.

The primary outcome of the study was patient decision for or against genetic screening as reported to the health care provider during the conversation. Secondary outcomes included whether a health care provider offered genetic screening and whether a patient ultimately underwent genetic screening as determined by medical record review. Chi-squared, Kruskal-Wallis and Wilcoxon rank sum tests were used to determine the effect of demographic variables on patient screening choices and completion of screening tests. As the number of visits per health care provider varied, the effects of College-recommend discussion topics and qualitatively-identified conversation themes was determined using logistic regression with generalized estimating equations to account for the clustering effect of multiple visits per health care provider. Qualitative coding was managed and organized using ATLAS.ti 4.2. Statistical analysis was performed with StataSE 13.1.

Results

We reviewed the first 267 visits in the larger study to find the first 200 visits meeting inclusion criteria for this analysis. Two patients were excluded due to unintelligible audio

recordings, one patient was excluded due to having previously completed first trimester screening at an outside institution, and 64 were excluded due to initiating prenatal care after 20 weeks gestation. The final analysis included 210 patients and 45 health care providers. Patient characteristics are summarized in table 1. Health care providers in our sample included 31 resident physicians, 5 certified nurse midwives, 7 nurse practitioners and 2 physician faculty. Each health care provider had an average of 4.7 study visits included in the analysis (range 1–10 visits). The majority of health care providers were Caucasian (87%) and female (92%). In multivariable model using GEE to control for clustering effects of health care providers, no significant difference was found between health care provider type, race or gender in offering genetic screening.

Most patients (190, 90%) were offered genetic screening at the visit. Of women offered screening, 148 (78%) decided for screening, 22 (12%) decided against screening and 20 (11%) were uncertain. Of the total sample, 149 women (71%) ultimately completed genetic screening tests. Seventy-three women (38%) were offered diagnostic testing, of whom 65 declined and 8 were uncertain. Nine women (4%) ultimately completed diagnostic testing, but six of these were prompted by abnormal ultrasound findings later in the pregnancy rather than the initial screen. No women were offered cell-free fetal DNA screening as an initial screening test. Average visit length was 29.4 minutes for visits in which health care providers offered genetic screening, and 38.7 minutes when genetic screening was not offered. Conversations about screening for fetal aneuploidy lasted 1.5 minutes on average (range 0.12 to 7.05 minutes).

The effects of patient characteristics on screening is shown in table 1. Women with more than two prior deliveries were less likely to complete screening than nulliparous women or women with two or fewer deliveries (P=0.01). Women who had prenatal genetic screening had significantly more prenatal visits than women who did not (11 vs 9, P=0.02). Maternal age, race, income, education and marital status were not associated with decision for or completion of screening tests for fetal aneuploidy.

Kappa inter-rater reliabilities were 0.72 for discussion of advantages and 0.91 for the availability of diagnostic testing. Detection rates and false positive rates were discussed so rarely that neither was seen in the recoded sample and thus kappas could not be calculated.Inter-rater reliability for discussion of disadvantages and limitations was low, but after discussion between authors this was determined to be due to difficulties in determining what constituted a "disadvantage" versus a "limitation" of genetic screening. When these two categories were collapsed into a single combined category and transcripts recoded accordingly the inter-rater reliability was 0.74. Inter-rater reliability for codes developed during thematic analysis (see table 2) was 0.62–0.93.

Adherence to College recommendations for prenatal genetic screening counseling was low, with health care providers mentioning all of the College-recommended topics in only 1.1% of visits. Inclusion of more College-recommended topics did not affect women's screening choices (Table 2). Conversations including two or more College-recommended topics were significantly longer than those including less than two topics (2.1 ± 1.2 minutes versus 0.9 ± 0.7 minutes, P<0.001) Health care providers discussed at least one advantage of

screening in 57% of conversations. Specific advantages reviewed included that first trimester and multiple marker screening are noninvasive, that patients could gain information about their baby's risk of aneuploidy, and that (for first trimester screening) testing involved an ultrasound which would let them see the baby and confirm pregnancy dating. Disadvantages or limitations were discussed in 50% of conversations, and included that screening tests were not diagnostic, that many birth defects are not detectable by first trimester screening, that diagnostic testing carries risk of miscarriage, that false-positive testing may increase patient anxiety, that testing must be performed during a narrow gestational age window.

Our qualitative exploration of prenatal genetic screening discussions showed that health care providers varied widely in their counseling style. Health care providers initiated the vast majority (99.5%) of these conversations, and had a relatively consistent "script" that they used with every patient in each of their recorded visits (see Appendix 1, available online at http://links.lww.com/xxx, for examples of health care provider scripts). Health care providers rarely (5% of conversations) strongly recommended screening. Patients asked questions about screening in only 26% of visits. Most patient questions focused on testing procedures and scheduling (e.g. "I have to get bloodwork too?" or "Will that be done today?"). A few patients asked questions clarifying the type of results (e.g. "But does that test show guaranteed that that's what it is?") or clarifying the difference between screening and invasive testing (e.g. "So it's not poking my belly is it?").

Health care provider discussions varied widely, from brief descriptions of the screening process to comprehensive discussions including explanations that the results of prenatal genetic screening tests were not diagnostic, the availability of amniocentesis or other tests to confirm the diagnosis of Down Syndrome, and the option of pregnancy termination for positive test results.

An example of a brief discussion is:

Health care provider: At this time in pregnancy ... they will be able to do what we call a first trimester screen. Which is a combination of an ultrasound that measures the back of the baby's neck, like a little skin fold, as well as a blood test that can see whether or not your pregnancy would be at increased risk for something like Down Syndrome. Would you be interested in something like that?

Patient: Yeah

Health care provider: OK. Um, does anything else run in your family history?

The following is an example of this more comprehensive counseling:

Health care provider: The last question, and this is something that's totally persondependent ... sometime between 11 ... and 13 weeks ... we offer women a first trimester screen. First trimester screening basically looks at the thickness of the baby's neck and takes bloodwork from you. And from those two things they can give you a prediction of your risk of having a baby with Down Syndrome.

Patient: OK

Health care provider: There are two scenarios where a woman may potentially want this. One, it may alter their decision to carry the pregnancy so they may wish to terminate the pregnancy if they found the baby had Down Syndrome ... the other reason some women would like it is just to know. And that's reason enough to have that type of study done. It's not invasive. If they find something positive they would offer you an amniocentesis or another form of definitive testing if you were higher risk.

Patient: OK

Health care provider: But if your risk is low, then that's all there is to it ... It's not like a yes or no thing ... I had a patient last week her risk was 1 in 11,000 ... your risk is low from the start. Because you're young, you've never had a history of anything like that before ... Women who tend to be higher risk are women over age 35. That's when the risk starts to go up much more.

Patient: I mean is it recommended for people who are my age? Is it just peace of mind sort of?

Health care provider: It's totally just personal preference.

Health care providers used a variety of techniques to describe the type of results obtained by prenatal genetic screening tests. Of those visits in which providers explained that the results of screening tests were not diagnostic, 67% (66 out of 99 conversations) used generic "high or low risk" terminology (eg, "So it doesn't say yes or no your baby does or doesn't have this disease, but it lets us know whether there's a[n] increased risk of your baby having it.") In two percent of visits, providers described results as a "percentage chance" of the baby having Down Syndrome, and in 31% of visits, providers described results as a probability (e.g. "It can be like 1 in 50 or maybe 1 in 50,000.") Health care providers describing results this way frequently used a mother's baseline risk of having a baby with Down Syndrome as a comparison to illustrate what a probability risk meant, as demonstrated in the following example:

Health care provider: You are young, you are twenty, right?

Patient: Yeah.

Health care provider: So your risk of having a baby with Down Syndrome is much less than even 1 in 1,000. Much less than that. But doing this test, it would say 'Wow, your risk is now 1 in 10,000 so much lower, or now it is 1 in 100 or 1 in 10.'

Not all providers clarified that prenatal genetic screening tests were not diagnostic of a baby having or not having Down Syndrome. In over half (51%) of these discussions, health care providers either did not discuss the topic at all, or referred to testing as "ruling out" Down Syndrome or "making sure the baby doesn't have [Down Syndrome]". Some of these providers introduced the testing as a screen to "see if your baby is at risk for Down Syndrome" without clarifying what "at risk" meant.

As opposed to discussions of routine prenatal laboratory work where health care providers strongly recommended testing, providers frequently (45% of conversations) emphasized that

prenatal genetic screening tests were optional (e.g. "We can talk about what if any genetic testing you are interested in. You do have some options depending on how much information you want. Those are optional. Not mandatory"). Many health care providers (37% of visits) then explained how a patient might use the information gained from prenatal genetic screening:

Health care provider: Reasons why you would choose to do it – one, in case you would terminate the pregnancy if you knew the baby would be challenged. Or two, that you wanted to know so you could do lots of preparation. If you feel that those don't apply to you or you wouldn't want to do any more testing because there is a little bit of a risk of miscarriage, you could choose not to do the testing.

Health care providers frequently discussed choices made by "some women" or hypothetical patients to review possible responses to screening results and to normalize a patient's feelings about whether or not she wanted prenatal genetic screening:

Health care provider: Some people really want to know anything and everything that could be wrong. Some don't. Some don't want to know any of that. It is not going to change the way they feel about the pregnancy and you know, sometimes it will make them more nervous than anything else.

In our analysis, the conversational techniques of emphasizing that screening is a personal choice, discussing how a patient might use screening results and normalizing patient's feelings about prenatal genetic screening were often used in tandem by providers.

Patients verbalized a variety of reasons for deciding for or against testing. Patients who chose to have screening wanted to "make sure the baby's healthy", to reassure themselves because of a family history of mental retardation, and to see the baby on the ultrasound (for first trimester screening). Women who declined testing cited additional anxiety provoked by testing (e.g. "It just adds too much stress", "I'd rather not worry about it while I'm pregnant"), prior poor experiences with testing, and that they would not terminate a pregnancy based on genetic testing results. Patients also reported considerable anxiety about the idea of diagnostic testing. Many women had heard about testing from friends or family members (e.g. "My cousin was telling me that when she was pregnant with her son the doctor at the hospital she was at, they were going to stick something through her belly button"). No women in the study chose invasive testing as a first line test, although some women voiced that they would consider it if screening was abnormal.

Discussion

We found that the majority of health care providers offered, and the majority of patients opted for prenatal genetic screening. Conversations ranged from brief mentions of screening to extensive discussion of all College-recommended discussion topics. While health care providers offered screening at the majority of visits, the lack of in-depth discussions by many health care providers raises concerns that health care providers may not be obtaining truly informed consent.

Prior surveys of physicians have reported that 90–100% of obstetric health care providers offer screening for fetal aneuploidy to their patients, which is consistent with our study results.^{14–16} However, other studies of patient understanding of prenatal genetic screening raise concerns about the content of health care provider counseling and patient comprehension.^{5,8–10,17} A qualitative study of 40 pregnant women with abnormal multiple marker screens found that 63% had no clear understanding of the screening test they had completed and thus had no idea how to interpret an abnormal result.⁵ A third-trimester survey of 710 women who had completed genetic screening earlier in the pregnancy showed that only 76% recalled correctly that prenatal genetic screening was an optional test, and only 88% recalled having completed a screening test at all.¹⁸

The lack of patient understanding of screening options may be due in part to inadequate health care provider counseling. While health care providers offered screening at 90% of visits, few included all College-recommended topics in their counseling. In 1998, Bernhardt observed health care providers counseling patients about multiple marker screening and found that while 81% of health care providers discussed the screening process, only 20–40% discussed the limitations of testing, the option of diagnostic testing, or the option of pregnancy termination for positive results.⁶ Even after two decades of experience with prenatal genetic screening our study found remarkably similar results.

Our finding that discussion of additional College-recommended topics does not influence women's screening choices is consistent with a recent finding by Kuppermann et al that use of an interactive decision-support tool decreased screening uptake.¹⁹ It is possible that instead of increasing screening uptake, comprehensive counseling instead allows women to make choices more consistent with their own values and beliefs, which may result in lower screening rates in populations where termination of pregnancy for anomalies is not considered appropriate.

One reassuring finding was the general use of a nondirective approach to screening conversations. Assisting women in making autonomous choices is one of the central tenants of genetic counseling.^{4,20–23} Health care providers accomplished this goal by emphasizing that screening is optional, and by discussing the choices made by other women to normalize a patient's screening choices. We were further encouraged that the duration of visits was not lengthened by discussions of prenatal genetic screening, and that even conversations including more College-recommended topics had an average duration of 2.1 minutes (compared to 0.9 minutes for inclusion of <2 topics).

One limitation of our study is the lack of discussion of cell-free fetal DNA screening by our providers. Our institution does not offer cell-free fetal DNA screening as a routine screening option for low-risk patients, and all patients who are candidates for cell-free fetal DNA screening are first referred to genetic counseling. Cell-free fetal DNA screening was first offered at our institution in March of 2012, and health care provider uptake of this option as a first-line screening for high-risk women has increased gradually over time since that point. Thus it is not surprising that none of our health care providers offered cell-free fetal DNA screening as a first line test. A further limitation of the study is the possibility that health care providers and patients modified their behavior due to the study recording. While the

participants and health care providers were unaware of the topic under study, it remains possible that conversations were more detailed (for the health care provider's part) or less forthright (for the patient's part) than they otherwise would have been without observation.

The major strengths of our study include the large sample size, the direct observation of visits rather than reliance on surveys or interviews, and the application of both quantitative and qualitative analysis. However, because all of our observations were conducted at a single academic center with primarily resident and midlevel health care providers, our findings may be influenced by our predominantly urban population and provider distribution.

Our study shows that comprehensive counseling about prenatal genetic screening is possible at first prenatal visits. While some health care providers kept to brief conversation scripts, many providers were able to discuss advantages and disadvantages of screening, review the type of results obtained by screening and how a patient might use them, discuss diagnostic testing options and encourage women to make autonomous choices all without overly lengthening the visit.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

Acknowledgments

Supported by the National Institute on Drug Abuse R01 DA026410 (Judy C. Chang, PI). This publication was made possible by Grant Number 5UL1 RR024153-05 from the National Center for Research Resources (NCRR), a component of the National Institutes of Health (NIH), and NIH Roadmap for Medical Research. Its contents are solely the responsibility of the authors and do not necessarily represent the official view of NCRR or NIH. Information on NCRR is available at http://www.ncrr.nih.gov/. Information on Re-engineering the Clinical Research Enterprise can be obtained from http://nihroadmap.nih.gov/clinicalresearch/overview-translational.asp.

References

- Parker SE, Mai CT, Canfield MA, Rickard R, Wang, Yi, Meyer RE, et al. Updated national birth prevalence estimates for selected birth defects in the United States, 2004–2006. Birth Defects Res A Clin Mol Teratol. 2010; 88:1008–1016. [PubMed: 20878909]
- ACOG Committee on Practice Bulletins. ACOG Practice Bulletin No 77: Screening for fetal chromosomal abnormalities. Obstet Gynecol. 2007; 109:217–227. [PubMed: 17197615]
- Society for Maternal-Fetal Medicine (SMFM). SMFM Consult Series #36: Prenatal aneuploidy screening using cell free DNA. Am J Obstet Gynecol. 2015; doi: 10.1016/j.ajog.2015.03.043
- Chervenak FA, McCullough LB, Sharma G, Davis J, Gross S. Enhancing patient autonomy with risk assessment and invasive diagnosis: an ethical solution to a clinical challenge. Am J Obstet Gynecol. 2008; 199:19e1–19.e4. [PubMed: 18355783]
- Hunt LM, de Voogd KB, Castañeda H. The routine and the traumatic in prenatal genetic diagnosis: does clinical information inform patient decision-making? Patient Educ Couns. 2005; 56:302–312. [PubMed: 15721973]
- Bernhardt BA, Geller GS, Doksum T, Larson SM, Roter D, Holtzman NA. Prenatal genetic testing: content of discussions between obstetric providers and pregnant women. Obstet Gynecol. 1998; 91:648–655. [PubMed: 9572205]
- 7. Smith DK, Slack J, Shaw RW, Marteau TM. Lack of knowledge in health professionals: a barrier to providing information to patients? Qual Health Care. 1994; 3:75–78. [PubMed: 10137588]
- Smith DK, Shaw RW, Marteau TM. Informed consent to undergo serum screening for Down's syndrome: the gap between policy and practice. BMJ. 1994; 309:776. [PubMed: 7950565]

- Van den Berg M, Timmermans DR, Ten Kate LP, Van Vugt JM, Van der Wal G. Are pregnant women making informed choices about prenatal screening? Genet Med. 2005; 7:332–338. [PubMed: 15915085]
- Moyer A, Brown B, Gates E, Daniels M, Brown HD, Kuppermann M. Decisions about prenatal testing for chromosomal disorders: perceptions of a diverse group of pregnant women. J Womens Health Gend Based Med. 1999; 8:521–531. [PubMed: 10839707]
- Chang JC, Alexander SC, Holland CL, Arnold RM, Landsittel D, Tulsky JA, et al. Smoking is bad for babies: obstetric care providers' use of best practice smoking cessation counseling techniques. Am J Health Promot. 2013; 27:170–176. [PubMed: 23286593]
- Demirci JR, Bogen DL, Holland C, Tarr JA, Rubio D, Li J, et al. Characteristics of breastfeeding discussions at the initial prenatal visit. Obstet Gynecol. 2013; 122:1263–1270. [PubMed: 24201684]
- Chang JC, Holland CL, Tarr JA, Rubio D, Rodriguez KL, Kraemer KL, et al. Perinatal illicit drug and marijuana use: an observational study examining prevalence, screening, and disclosure. Am J Health Promot. 2015; doi: 10.4278/ajhp.141215-QUAL-625
- Green JM. Serum screening for Down's syndrome: experiences of obstetricians in England and Wales. BMJ. 1994; 309:769–772. [PubMed: 7950560]
- Press N, Browner CH. Why women say yes to prenatal diagnosis. Soc Sci Med. 1997; 45:979–989. [PubMed: 9257391]
- Driscoll D, Morgan M, Schulkin J. Screening for Down syndrome: changing practice of obstetricians. Am J Obstet Gynecol. 2009; 200:459e1–459.e9. [PubMed: 19318157]
- Marteau TM, Dormandy E. Facilitating informed choice in prenatal testing: how well are we doing? Am J Med Genet. 2001; 106:185–190. [PubMed: 11778978]
- Bryant AS, Norton ME, Nakagawa S, Bishop JT, Pena S, Gregorich SE, et al. Variation in Women's Understanding of Perinatal Testing. Obstet Gynecol. 2015; 125:1306–1312. [PubMed: 26000501]
- Kupperman M, Pena S, Bishop JT, Nakagawa S, Gregorich SE, Sit A, et al. Effect of enhanced information, values clarification, and removal of financial barriers on use of prenatal genetic testing: a randomized clinical trial. JAMA. 2014; 312:1210–1217. [PubMed: 25247517]
- Chervenak FA, McCullough LB. Ethical issues in perinatal genetics. Semin Fetal Neonatal Med. 2011; 16:70–73. [PubMed: 21051301]
- Kuppermann M, Learman L, Gates E, Gergorich SE, Nease RF, Lewis J, et al. Beyond race or ethnicity and socioeconomic status: predictors of prenatal testing for Down syndrome. Obstet Gynecol. 2006; 107:1087–1097. [PubMed: 16648415]
- 22. Van den Berg M, Timmermans DR, Kleinveld JH, Van Eijk JT, Knol KL, Van der Wal G, et al. Are counsellors' attitudes influencing pregnant women's attitudes and decisions on prenatal screening? Prenat Diagn. 2007; 27:518–524. [PubMed: 17367105]
- Hertig SG, Cavalli S, Burton-Jeangros C, Elger BS. 'Doctor, what would you do in my position?' Health professionals and the decision-making process in pregnancy monitoring. J Med Ethics. 2014; 40:310–314. [PubMed: 23543803]
- Norton, ME. Counseling for Prenatal Aneuploidy Screening; Obstet Gynecol. 2013. p. 122Podcast available online at: http://journals.lww.com/greenjournal/pages/podcastepisodes.aspx?podcastid=4

Box 1

Suggested Prenatal Genetic Screening Counseling

The American College of Obstetricians and Gynecologists recommends providing information about the detection and false positive rates, advantages, disadvantages and limitations of screening tests as well as the risks and benefits of diagnostic procedures to patients so that they can make informed decisions.^{2,24} One possible framework for this discussion includes:

- Discuss the screening process including the need for ultrasound and blood testing (for first trimester screening) or blood testing alone (for multiple marker screening)
- Review that the test screens for Down syndrome and Trisomy 18 (and open neural tube defects if the multiple marker screen is offered) but does not test for other birth defects or causes of developmental delay.
- Clarify the difference between screening tests and diagnostic tests. Discuss the type of results obtained from screening tests and the option of diagnostic testing either as a first-line or confirmatory test.
- Emphasize that testing is optional, encourage patients to reflect on their own values and beliefs regarding screening.
- Encourage patients to consider how they would respond to positive test results:
 - Invasive diagnostic testing?
 - Termination of pregnancy if positive results are confirmed?
 - Preparation of family and health providers to care for a child with special needs?
- Provide referral to genetic counseling if patients have further questions.

Author Manuscript

Author Manuscript

Author Manuscript

Table 1

Baseline Maternal Characteristics by Decision for and Completion of Screening Tests for Fetal Aneuploidy

Patient Characteristic	Decision For or Again	For or Against Genetic Screening at the Time of Visit *	time of Visit*		Completion of Screening Test †	st ŕ	
	Decided for (n=148)	Decided against (n=22)	Undecided (n=20)	d	Had Screening Test (n=149)	Did Not Have Screening Test (n=61)	Ч
Age (years)	24.9 ± 5.1	24.9 ± 5.9	28.9±8.2	0.12	24.9±5.5	26.2±6.0	0.11
Parity							
0	71 (48.0)	7 (31.8)	10 (50)	0.02	69 (46.3)	23 (37.7)	0.01
1–2	64 (43.2)	8 (36.4)	10 (50)		69 (46.3)	24 (39.3)	
More than 2	13 (8.8)	7 (31.8)	0		11 (7.4)	14 (23.0)	
Gestational age at first prenatal visit (weeks)	9.7±3.4	11.0 ± 4.4	9.4±4.1	0.34	9.8±3.5	9.6±4.0	.17
Number of prenatal appointments \sharp	11 (3–19)	8.5 (2–15)	11 (6–15)	0.17	11 (2–19)	9 (2–17)	0.02
Race							
African American	85 (57.4)	10 (45.5)	9 (45.0)	0.14	82 (55.0)	33 (54.1)	0.71
Caucasian	49 (33.1)	6 (27.3)	7 (35.0)		51 (34.2)	19 (31.2)	
Other	14 (9.5)	6 (27.3)	4 (20.0)		16 (10.7)	9 (14.8)	
Education high school or less	71 (48.0)	12 (54.5)	9 (45.0)	0.80	73 (49.0)	34 (55.7)	0.38
Income \$15000 or less annually	89 (60.5)	16 (72.7)	12 (60.0)	0.54	95 (63.8)	35 (58.3)	0.46
Married or cohabitating with partner	76 (51.4)	13 (59.1)	13 (65.0)	0.45	79 (53.0)	30 (50.0)	0.69
Data are mean+SD. n (%) or median(range) unless otherwise snecified	ess otherwise specified						

Data are mean±SD, n (%) or median(range) unless otherwise specified

Obstet Gynecol. Author manuscript; available in PMC 2017 June 01.

P values based on ttest, Kruskal-Wallis or Wilcoxon rank sum test

* out of 190 patients offered screening by providers

 $\overset{\tau}{ }^{}_{ }$ out of total sample of 210 patients

 t^{\dagger} excluding women whose pregnancies ended in termination, miscarriage, or unknown status

Author Manuscript

Table 2

The Effects of Inclusion of American College of Obstetricians and Gynecologists-Recommended Counseling Elements and Other Elements of Provider Counseling on Maternal Decision for and Completion of Screening Tests for Fetal Aneuploidy

Counseling Element	Conversations Including Counseling Element (n=190)	Patients Choosing Screening After Counseling	\mathbf{P}_{*}^{*}	Patients Completing Screening Test After Counseling	4
Counseling Elements Recommended by ACOG:					
Detection Rate	2 (1.1)	2/2 (100)	1.0	2/2 (100)	1.0
False Positive Rate	3 (1.6)	2/3 (66.6)	0.56	2/3 (66.6)	0.82
Advantages	104 (54.7)	84/104 (80.8)	0.19	77/104 (74.0)	0.62
Disadvantages or Limitations	92 (48.7)	69/92 (75.0)	0.87	67/92 (72.8)	0.89
Availability of Diagnostic Testing	73 (35.3)	53/73 (72.6)	0.27	52/73 (71.2)	0.96
Reviews the screening process	163 (85.8)	130/163 (79.8)	0.049	118/163 (72.4)	0.87
Gives a woman's baseline (age-based) risk	55 (29.1)	39/55 (70.9)	0.07	42/55 (76.4)	0.42
Explains what is Down Syndrome	38 (20.0)	26/38 (68.4)	0.35	26/38 (68.4)	0.51
Explains that screening tests also screen for Trisomy 18	49 (25.8)	40/49 (81.6)	0.34	39/49 (79.6)	0.21
Explains that screening results are not diagnostic	97 (51.1)	73/97 (75.3)	0.86	72/97 (74.2)	0.58
Emphasizes that screening is optional	85 (44.7)	56/85 (65.9)	0.002	58/85 (68.2)	0.20
Discusses the option of pregnancy termination if results are abnormal	31 (16.3)	22/31 (71.0)	0.29	19/31 (61.3)	0.13
Discusses how screening results might be useful to a patient (reassurance, diagnostic testing, termination, etc.)	71 (37.4)	54/71 (76.1)	0.75	52/71 (73.2)	0.88

Obstet Gynecol. Author manuscript; available in PMC 2017 June 01.

Patients who were undecided at the time of visit included with patients who declined screening in this analysis

Data are n (%) or n/n (%)

P values based on logistic regression with generalized estimating equations