First-Trimester Aneuploidy Risk Assessment: The Impact of Comprehensive Counseling and Same-Day Results on Patient Satisfaction, Anxiety, and Knowledge

Nathan S. Fox, M.D.,[1] Andrei Rebarber, M.D.,[1] Chad K. Klauser, M.D.,[1] Ashley S. Roman, M.D., M.P.H.,[1] and Daniel H. Saltzman, M.D.[1]

ABSTRACT

We evaluated the added benefit of a comprehensive counseling protocol for first-trimester aneuploidy risk assessment. We performed a prospective cohort study surveying patients referred for first-trimester aneuploidy risk assessment. We compared responses between women who underwent serum testing done in advance of their ultrasound such that their final risk assessment was given to them the same day as their ultrasound (comprehensive) versus women who underwent serum testing the same day as their ultrasound and who therefore received their final risk assessment later (standard). Response rate was 94.8%. The comprehensive group was significantly more likely to receive counseling in accordance with recommended American College of Obstetricians and Gynecologists (ACOG) guidelines, had significantly greater reduction in anxiety and increased satisfaction, and was more likely to report an increased understanding of their results. The comprehensive group scored significantly higher on test-style questions about aneuploidy risk assessment. Comprehensive aneuploidy risk assessment counseling including same-day results is associated with increased patient understanding and satisfaction, decreased anxiety, and increased adherence to ACOG guidelines.

KEYWORDS: Aneuploidy, risk assessment, nuchal, Down's, counseling

The American College of Obstetricians and Gynecologists (ACOG) supports first-trimester risk assessment for fetal aneuploidy in the general population.[1] The combination of first-trimester ultrasound for nuchal translucency measurement, serum pregnancy associated plasma protein A, and free β-human chorionic gonadotropin (combined risk assessment) is a reliable test for aneuploidy risk assessment.[2–4] The administration of the first-trimester combined risk assessment tests can be done in several different ways. The serum tests can be drawn the same day as the ultrasound, in which case the final risk assessment results will be available within a week. Alternatively, the serum tests can be drawn in advance of the ultrasound, so that the final risk assessment results are available immediately after the ultrasound is completed. Some centers offer point of care testing such that the serum tests are drawn on the same day as the ultrasound, and results are available within a few hours, but this is not currently available in the United States.[2] In one survey in the United Kingdom, when over

[1]Carnegie Imaging for Women, New York, New York.

Address for correspondence and reprint requests: Nathan S. Fox, M.D., Carnegie Imaging for Women, 1245 Madison Avenue, New York, NY 10128 (e-mail: nfox@mfmnyc.com).

Received: February 15, 2010. Accepted after revision: May 10, 2010.

DOI: http://dx.doi.org/10.1055/s-0030-1262506.

ISSN 0735-1631.

1000 patients were asked to select among five hypothetical choices for aneuploidy risk assessment delivery, point of care testing was the preferred method, indicating the desire of patients to have their results as soon as possible.[5] Because this was only a hypothetical choice, this study could not assess whether patient satisfaction is actually increased with one delivery method compared with another. Additionally, patient satisfaction is only one clinical outcome that should be considered when delivering aneuploidy risk assessment. Administering the test in a manner such that the patient can understand the test and its results is also necessary in providing proper counseling. Additionally, because most people will receive reassuring results, the method should ideally attempt to decrease anxiety for most patients. Despite multiple national and international organizations establishing comprehensive first-trimester screening guidelines, there are limited data comparing different methods currently available used to procure the test.

In our ultrasound unit, we offer two options for delivery of first-trimester risk assessment. One option is to have the serum tests drawn in advance of the ultrasound. These patients are given their final risk assessment results the same day as their ultrasound by one of our maternal fetal medicine (MFM) specialists. The other option is to have the serum tests drawn the same day as the ultrasound. These patients receive their final risk assessment results from their referring physicians, usually 1 week later. The purpose of this study was to compare patient satisfaction with, and patient understanding about, aneuploidy risk assessment in regards to these two methods.

METHODS

This is a prospective cohort study of patients undergoing first-trimester aneuploidy risk assessment in a single ultrasound unit. The ultrasound unit serves as a referral center for several local obstetricians delivering at different hospitals in New York City. Patients referred to our unit have the option of having their serum tests drawn in advance of their ultrasound, either in our unit, at home, or in their referring obstetrician's office. Serum testing in all three locations is obtained with a finger-stick blood sample that is blotted onto a dedicated laboratory card that is mailed to the risk assessment laboratory (NTD Laboratories, Melville, NY). For these patients, when we have completed their first-trimester risk assessment ultrasound, we input the ultrasound data (crown-rump length, nuchal translucency measurement, nasal bone evaluation) into an online program designed by our risk assessment laboratory (NTD Laboratories). This online program combines the previously drawn serum results with the newly inputted ultrasound data and instantly returns a final risk assessment. The MFM specialist interpreting the images formally counsels the patient at that time regarding this final combined aneuploidy risk assessment. The counseling session performed by one of four MFM specialists in our group consisted of an attempt to strictly adhere to ACOG guidelines such that risk assessment results were presented in a numerical fashion, patients were provided the opportunity to ask questions regarding the testing, and options for invasive procedures (chorionic villus sampling and/or amniocentesis) was provided regardless of test results or patient age.

Alternatively, patients can choose to have their serum tests drawn on the same day as their ultrasound. For these patients, the ultrasound data are mailed to the laboratory along with the serum sample, and the final results are returned to the referring obstetrician, usually within 1 week. The qualitative results of the ultrasound are discussed with these patients, but these patients are only given formal aneuploidy counseling by one of our MFM specialists if an abnormality is seen on ultrasound or if the nuchal translucency measures greater than or equal to 3 mm. Otherwise, all counseling is done by their referring obstetricians after the final quantitative results are available.

This study compares two cohorts: patients who had their serum tests drawn in advance of their ultrasound such that they received aneuploidy risk assessment counseling in our unit on the day of their ultrasound (comprehensive group) and patients who had their serum tests drawn on the same day as their ultrasound such that they received aneuploidy risk assessment counseling from their referring obstetrician at a later time (standard group).

All of our referring obstetricians are made aware of both options for the delivery of aneuploidy risk assessment. All pretest counseling was performed by the referring obstetricians, and the decision which option to choose was made by either the patients and/or their referring obstetricians. After Institutional Review Board approval was obtained, we conducted a questionnaire-based survey of patients in our ultrasound unit from April to July 2009. We surveyed all patients who returned to our unit anytime between 16 and 24 weeks for another ultrasound. At the time of the 16- to 24-week ultrasound, these patients were given an anonymous questionnaire to complete by the clerical staff and advised to return them to an anonymous collection box placed in the ultrasound waiting room. We only included patients who were referred to our unit. Any patient who received prenatal care from any of the MFM specialists in our unit was excluded and not given a questionnaire.

The survey was designed to elicit patient satisfaction with, and patient understanding of, aneuploidy risk assessment. Additionally, we examined whether certain ACOG recommendations regarding aneuploidy risk assessment[1] were being followed, including discussing the difference between screening and invasive diagnostic

Table 1 Baseline Characteristics

	Standard Group ($n = 186$)	Comprehensive Group ($n = 193$)	p
Mean age (y)	32.91 ± 4.32	33.98 ± 3.66	0.014
Age 35 or more	48/173 (27.7%)	74/192 (38.5%)	0.029
Race			
American Indian or Alaskan native	1/162 (0.6%)	1/174 (0.6%)	0.425
Asian or Pacific Islander	19/162 (11.7%)	23/174 (13.2%)	
Black	7/162 (4.3%)	4/174 (2.3%)	
Hispanic	16/162 (9.9%)	8/174 (4.6%)	
White	112/162 (69.1%)	131/174 (75.3%)	
Other	7/162 (4.3%)	7/174 (4.0%)	
Education—undergraduate or postgraduate degree	154/163 (94.5%)	175/175 (100%)	0.002
Prior children	64/161 (39.8%)	52/175 (29.7%)	0.053
Prior miscarriages	37/160 (23.1%)	49/173 (28.3%)	0.279
Prior elective termination of pregnancy	23/156 (14.7%)	30/169 (17.8%)	0.463
Prior aneuploidy risk assessment	106/161 (65.8%)	124/175 (70.9%)	0.323
In vitro fertilization	11/162 (6.8%)	8/175 (4.6%)	0.378
Baseline anxiety—moderate or significant (%)*	63/181 (34.8%)	84/191 (44%)	0.071

*Based on answer to the following multiple choice question: ''Before you underwent the Down's syndrome screening, how would you describe your level of anxiety about your fetus having or not having an abnormality? (A) No anxiety at all; (B) Minimal anxiety; (C) Moderate anxiety; (D) Significant anxiety.''

testing, offering invasive diagnostic testing to all patients regardless of age, and reporting the results as a numerical risk (1 in n), as opposed to normal/ abnormal or high risk/low risk.

Because this was an anonymous questionnaire, we could not ascertain the actual risk assessment results for each patient in our study. Therefore, this information could not be factored into the analysis.

We received no corporate sponsorship/support regarding this study development and performance. Chi-square test, Student t test, and Mann-Whitney U test were used when appropriate (SPSS for Windows 16.0, 2007; Chicago, IL). A p value of <0.05 was considered significant. Responses left blank were censored.

RESULTS

A total of 400 questionnaires were distributed, and 379 (94.8%) were completed and returned. The patients were divided almost evenly between the two groups: 193 (50.9%) patients were in the comprehensive group and 186 (49.1%) patients were in the standard group. Baseline characteristics are described in Table 1. The patients in the comprehensive group were slightly older. Overall, our patient population was highly educated, with 94.5% and 100% of patients having completed at least an undergraduate education in the standard and comprehensive groups, respectively ($p = 0.002$). One hundred percent of patients enrolled had private health insurance.

Patients in the comprehensive group were significantly more likely to report receiving their counseling in adherence to ACOG recommendations; patients in the comprehensive group were significantly more likely to report receiving their risk as a numerical value and to report being informed that an invasive procedure is available, if desired (Table 2). Additionally, there were significantly more patients in the comprehensive group who reported that the results of their aneuploidy risk assessment reduced their level of anxiety (Table 2). Seven percent of patients in the standard group indicated that no one reviewed their risk assessment results with them at any time, compared with no patients in the comprehensive group ($p < 0.001$).

Table 3 describes patient's responses to questions with a 1 to 5 answer scale (1 = strongly disagree;

Table 2 Survey Responses Based on Type of Counseling Protocol

	Standard Group ($n = 186$)	Comprehensive Group ($n = 193$)	p
When giving me the results, I was told that the results were a risk of 1 in something (1 in 5000 for example).	79/178 (44.4%)	135/189 (71.4%)	<0.001
When receiving the results, I was told that an invasive procedure (chorionic villus sampling or amniocentesis) is available if desired.	64/173 (37%)	142/187 (75.9%)	<0.001
The results of my aneuploidy risk assessment reduced my level of anxiety.	116/169 (68.6%)	153/182 (84.1%)	<0.001

Table 3 Self-Reported Scores on a 1–5 Scale* Regarding Aneuploidy Risk Assessment, Based on Type of Counseling Protocol

	Standard Group ($n = 186$)	Comprehensive Group ($n = 193$)	p (Mann-Whitney U)
I am satisfied with the overall process of the test.	4 (4,5)	5 (4,5)	0.067
I am satisfied with the time it took to receive the final results of the test.	4 (3,5)	5 (4,5)	<0.001
I am satisfied with the way the results of the test were explained to me.	4 (3,5)	5 (4,5)	<0.001
I understand the results of my test.	4 (3,5)	5 (4,5)	<0.001
I am glad I underwent the test.	4 (4,5)	5 (4,5)	<0.001
I would recommend this test to a friend.	5 (3,5)	5 (4,5)	0.002
I understand the difference between the results of a screening test such as this test and a diagnostic test such as CVS or amniocentesis.	4 (2,5)	5 (4,5)	<0.001
The person giving me the results appropriately explained the risks of an invasive procedure (CVS, amniocentesis).	4 (2,5)	4 (3,5)	<0.001
The results of my own test were reassuring to me.	4 (3,5)	5 (4,5)	0.003

*1 = strongly disagree; 2 = disagree; 3 = neither agree nor disagree; 4 = agree; 5 = strongly agree.
Median (10,90). CVS, chorionic villus sampling.

2 = disagree; 3 = neither agree nor disagree; 4 = agree; 5 = strongly agree). Patient satisfaction was significantly higher in the comprehensive group compared with the standard group. Additionally, patients in the comprehensive group gave significantly higher ratings in regards to their understanding of the test, their ability to distinguish between a screening test and a diagnostic test, and how well the person giving the counseling explained the risks of invasive testing.

In addition to asking patients to self-report their level of understanding of aneuploidy risk assessment, patients were also asked to answer two questions designed to assess their actual understanding of aneuploidy risk assessment. Question 1 read: "Which of the following women has a higher risk of her fetus having Down's syndrome? (a) A 27-year-old woman whose results show a Down's syndrome risk of 1 in 5000. (b) A 37-year-old woman whose results show a Down's syndrome risk of 1 in 5000. (c) These two women have the same risk of having a fetus with Down's syndrome." There were significantly more patients in the comprehensive group who correctly answered "c" (65.4% versus 44.6%, $p < 0.001$).

Question two was a true/false question regarding the following statement: "Down's syndrome screening (blood tests plus ultrasound) can diagnose a fetus with Down's syndrome." There was a trend toward more women in the comprehensive group correctly answering this question "false" (70.6% versus 60.6%, $p = 0.051$).

DISCUSSION

In this study, a first-trimester aneuploidy risk assessment counseling protocol including same-day results and designated one-to-one counseling was significantly associated with increased patient satisfaction, reduced anxiety, and increased knowledge. It is not possible to discern which aspect of the counseling protocol was most effective, but it appears as if both the same-day results and the dedicated counseling were important. For example, patients reported increased satisfaction with the timing of their results when given same-day results. This agrees with a hypothetical survey in which patients indicated that receiving same-day results was the most appealing option to them.[5] The addition of a dedicated counseling session also appeared to be useful. Patients reported decreased anxiety and a better understanding of aneuploidy risk assessment, and they scored better on specific questions assessing their knowledge of aneuploidy risk assessment. Additionally, there appeared to better adherence to ACOG recommendations including reporting the risk as a numerical value, explaining the difference between screening and invasive diagnostic testing, and offering invasive diagnostic testing to all patients regardless of age.[1] Furthermore, 7% of patients in the standard group reported that no one reviewed their results with them, whereas no patients in the comprehensive group reported this. This difference alone underscores the importance of an organized delivery system for aneuploidy risk assessment.

Strengths of our study include a very high response rate of 94.8%. Our patients were also highly educated, making it unlikely that their scores on the knowledge-based questions were influenced by a lack of general education. Scores on knowledge-based questions may be lower in a less-educated population. This could make the dedicated counseling even more important in these populations.

Because we distributed the survey in the second trimester, any patient who had her first-trimester

aneuploidy risk assessment in our unit and did not return for a second-trimester ultrasound was not surveyed. In theory, this could have introduced some bias into the study. However, it would not have been possible to survey patients at the time of their aneuploidy risk assessment, as all patients having their serum tests drawn that day would not be reviewing their final results for another week. Additionally, because there were a similar number of patients in the standard and comprehensive groups, it is unlikely that this possible bias affected the study results.

Since January of 2007, ACOG issued practitioner guidelines recommending all pregnant patients presenting in the first trimester should be offered the option of first-trimester screening. Since that time, several companies have developed a myriad of different testing protocols for commercial use. Additionally, the procurement of testing in clinical practice has not been well defined nor has it been studied outside the context of previously published standardized trials. Additionally, in the United States, population administration of first-trimester screening has become decentralized from dedicated fetal testing centers traditionally used in European countries. This later issue, despite standardized training and certification to actually perform the test, may provide varying results on actual patient understanding, satisfaction, and even the availability of appropriate invasive testing.

In our experience, explaining aneuploidy risk assessment to patients and reviewing results of aneuploidy risk assessment with patients is time-consuming and requires a very good understanding of the nuances of the test. Our study indicates that having devoted time to review the results with someone very familiar with aneuploidy risk assessment is beneficial to patients. This should be considered when enacting policies and procedures in regards to the delivery of first-trimester aneuploidy risk assessment.

REFERENCES

1. American College of Obstetricians and Gynecologists. Screening for chromosomal abnormalities. ACOG Practice Bulletin 77. Washington, DC: ACOG; 2007
2. Bindra R, Heath V, Liao A, Spencer K, Nicolaides KH. One-stop clinic for assessment of risk for trisomy 21 at 11–14 weeks: a prospective study of 15 030 pregnancies. Ultrasound Obstet Gynecol 2002;20:219–225
3. Wapner R, Thom E, Simpson JL, et al. First-trimester screening for trisomies 21 and 18. N Engl J Med 2003;349: 1405–1413
4. Malone FD, Canick JA, Ball RH, et al. First-trimester or second-trimester screening, or both, for Down's syndrome. N Engl J Med 2005;353:2001–2011
5. Spencer K, Aitken D. Factors affecting women's preference for type of prenatal screening test for chromosomal anomalies. Ultrasound Obstet Gynecol 2004;24:735–739