Screening for Down syndrome: changing practice of obstetricians

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OBJECTIVE: We sought to assess the impact of American College of Obstetrician and Gynecologists (ACOG) guidelines on the practices and knowledge of obstetricians regarding screening for Down syndrome 1 year later.

STUDY DESIGN: A questionnaire on Down syndrome screening was mailed to 968 ACOG Fellows and Junior Fellows.

RESULTS: The response rate was 53%. The majority (95%) of respondents offer Down syndrome screening to all pregnant patients; 70% of general obstetricians offer the first-trimester screen and 86% the quad screen. Almost two-thirds (63%) of respondents are offering patients $\geq$ 1 combination of first- and second-trimester screening tests. For women aged $<$ 35 years, 70% offer amniocentesis selectively and 15% routinely. Chorionic villus sampling is offered less frequently. Respondents who more closely read the bulletin were more likely to say their practice had changed, answered more knowledge questions correctly, and felt more qualified to counsel patients. Most (85%) obstetricians personally counsel patients about Down syndrome risk and screening tests. The majority (94-95%) of respondents have access to adequate resources for screening within a 90-minute drive.

CONCLUSION: Obstetricians have adopted a new paradigm for Down syndrome screening. First-trimester screening has been incorporated into prenatal care. Experience with these current screening tests will likely influence future guidelines and challenge the long-standing tradition of offering diagnostic testing based on maternal age. This study highlights the need for concise, unambiguous guidelines and a need to address unresolved issues in Down syndrome screening.

Key words: Down syndrome, first-trimester screen, maternal serum screen, nuchal translucency, questionnaire

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The American College of Obstetricians and Gynecologists (ACOG) published a practice bulletin, *Screening for Fetal Chromosomal Abnormalities*, in January 2007.[1] The practice bulletin was developed by the ACOG Committees on Practice Bulletins–Obstetrics and Genetics, and the Society for Maternal Fetal Medicine (SMFM). One of the major objectives of this document was to provide obstetrician-gynecologists with a summary and evaluation of recent evidence for the use of biochemical and sonographic markers for Down syndrome screening.

The guidelines also offered a new paradigm for offering screening and diagnostic testing for Down syndrome. Improvements in the sensitivity of noninvasive first- and second-trimester screening tests for Down syndrome challenged the use of maternal age cutoffs to determine whether women should be offered invasive diagnostic testing or a screening test. The practice bulletin recommends that, ideally, all women regardless of maternal age should be offered aneuploidy screening before 20 weeks' gestation. The guidelines also acknowledge that the decision to screen or test for Down syndrome in a pregnancy is a personal one and if adequately informed of the risks, benefits, and limitations of both screening and diagnostic tests, patients should have the option to have a diagnostic test regardless of maternal age. Therefore, maternal age of $\geq$ 35 years should not be used to determine whether a woman is eligible for an amniocentesis or chorionic villus sampling (CVS). Last, in recognition of the complexity and limited availability of some screening and diagnostic tests at the time, the practice bulletin provides obstetricians with suggestions for implementing a Down syndrome screening strategy in their practices.

The objective of this study was to assess the impact of the ACOG guidelines on physicians' practice and knowledge regarding Down syndrome screening a year after the practice bulletin was published. The study was also designed to evaluate the availability and proximity of resources for genetic screening and testing, and estimate patients' acceptance of Down syndrome screening.

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Materials and Methods

The questionnaire was developed in consultation with obstetrician-geneticists and was pilot tested on a sample of practicing obstetrician-gynecologists before final distribution. The study was approved by the institutional review board at the University of Pennsylvania, Philadelphia, PA. Questionnaires were mailed in October 2007 to 968 Fellows and Junior Fellows of the ACOG who practice within the United States. At least 1 questionnaire was sent to every state of the United States except Alaska and Wyoming. Of these subjects, 477 were randomly selected from the Collaborative Ambulatory Research Network (CARN). Members of CARN are practicing obstetrician-gynecologists who have volunteered to participate in survey studies on a regular basis. CARN was established to facilitate assessment of clinical practice patterns and aid the development of educational materials.[2] The remaining 491 subjects consisted of a computer-generated random sample of ACOG Fellows and Junior Fellows who had not received a survey from ACOG during the previous 2 years (non-CARN). All nonrespondents received up to 4 mailings, each separated by a few weeks. Questionnaires returned by April 2008 were included in the study.

Survey responses were anonymous and recorded demographic details of physicians and their patient population and assessed practices and knowledge regarding Down syndrome screening, the availability of genetic counseling, ultrasonography, diagnostic tests, and abortion services. The questionnaire contained 51 questions, some multipart: 9 demographics questions, 31 practice questions, 6 knowledge questions, and 5 professional education questions. Question formats included multiple choice, yes/no, check all that apply, and fill in the blank. A brief survey on eating disorders was included in the same mailing. This resulted in a final document of 3 double-sided pages.

The data were analyzed using a personal computer-based software package (SPSS, version 16.0; SPSS Inc, Chicago, IL). Descriptive statistics were computed for the measures used in the analyses, which are reported as mean ± SEM. Student t test and analysis of variance were used to compare group means of continuous variables; where post hoc analyses were conducted, the Bonferroni correction was used. Differences on categorical measures were assessed using χ^2. Group differences on ordinal measures and knowledge scores were assessed using the Mann-Whitney U or Kruskal-Wallis test. Correlations used the Pearson r or Spearman ρ coefficient. All analyses were tested for significance using an α of 0.05.

Results

A total of 517 questionnaires were returned. Surveys from 10 respondents were judged invalid (physician retired, returned to sender, blank survey), resulting in a valid response rate of 53% (507/958). There were responding physicians from the District of Columbia and from every state except Alaska, Montana, and Wyoming. Respondents' mean age (49.35 ± .434 years) closely matched that of nonrespondents (49.35 ± .468 years). Female subjects were marginally more likely to respond than male subjects (56% of female subjects [247/439] responded vs 50% of male subjects [260/519]; $\chi^2 = 3.632$; $P = .057$). CARN members were significantly more likely to respond than non-CARN members (CARN = 61%, 289/471; non-CARN = 45%, 218/487; $\chi^2 = 24.47$; $P < .001$). CARN members were significantly but only somewhat more likely than non-CARN members to say they offer Down syndrome screening to all pregnant patients (97% vs 91%; $P < .05$) and were less likely to offer independent first- and second-trimester screen (14% vs 28%; $P < .05$), and serum integrated screen in a twin gestation (2% vs 7%; $P < .05$); data were collapsed across membership group on the remaining variables.

Analyses were limited to respondents who care for obstetric patients (n = 391; 77% of respondents) because we were interested in how obstetrician-gynecologists manage screening and diagnosis of Down syndrome during prenatal care. Table 1 summarizes the characteristics of respondents who treat obstetric patients. The majority (n = 351; 90%) of these reported a primary medical specialty of general obstetrics and gynecology (obstetrician-gynecologists). A total of 37 respondents self-identified as maternal-fetal medicine specialists (MFMs) and differed on a number of physician/patient characteristics (Table 1). More of the MFMs practice in an academic setting within urban areas in the Northeast. Preliminary analyses indicated substantial differences in their responses; therefore, the data were analyzed separately. Of the respondents who do not manage obstetric patients (23%; n = 115), most practice gynecology or a gynecologic subspecialty.

Screening for Down syndrome

Most (95%) responding physicians said they offer Down syndrome screening to all of their pregnant patients. Of the 5% who do not offer it to all pregnant patients, 18 or 95% said they routinely offer it to all pregnant patients aged ≥ 35 years. Table 2 lists several first- and second-trimester screening tests and the frequency with which respondents use them. In all, 68% of obstetrician-gynecologists and 75% of MFMs offer second-trimester quad screen (α-fetoprotein, β-human chorionic gonadotrophin [hCG], unconjugated estriol, inhibin-A) frequently, whereas the triple screen is used frequently by only 16% of obstetrician-gynecologists and 11% of MFMs.

Of the obstetrician-gynecologists, 42% offer the combined first-trimester screen (nuchal translucency [NT] measurement, pregnancy plasma protein-A [PAPP-A], β-hCG) frequently and 28% offer it case by case. Half (50%) of obstetrician-gynecologists typically offer the first-trimester screen as a stand-alone test without a follow-up second-trimester screening test; 33% to all patients who present for prenatal care before 14 weeks' gestation; and an additional 17% offer it in specific circumstances, including patient request, insurance reasons, multiple gestation, maternal age, the patient desires CVS, the patient would choose to terminate an affected pregnancy, or a history of high-risk pregnancy. Among MFMs the combined first-trimester screen was used fre-