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OPINION

Preconception counseling: do patients learn about genetics from their obstetrician gynecologists?

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Abstract
Purpose The purpose of this observational survey study is to assess genetic knowledge in reproductive-aged women and to determine the role played by their obstetricians in their education.
Methods A 31-item survey was distributed via an internet survey service to women between the ages of 18 and 45. The survey included subject demographics, a query regarding the source of subjects' knowledge of genetics, and 6 question genetics quiz with 3 fundamental questions and 3 advanced questions. Subjects were divided into parous and nulliparous groups, and responses were compared using student's *t*-test for continuous variables and chi square for proportions.
Results Participants included 207 parous and 221 nulliparous women. There were no differences in demographic characteristics including age and education. Parous women scored significantly higher than nulliparous women on the fundamental genetics quiz (71 vs 61 %, $p=0.03$). This difference remained but was no longer significant when the 3 advanced questions were included (48 vs 42 %). Only 39 % of parous and 8 % of nulliparous subjects listed their physician as one of their main sources of genetic information. 78 % of all subjects stated that they would prefer to receive genetic information from their physicians over other sources.
Conclusions Recently parous women scored higher on a genetics assessment quiz than did their nulliparous counterparts, but the majority did not cite their obstetrician gynecologists as a main source of information. As genetic counseling and testing are becoming increasingly important aspects of obstetrical care, obstetricians should play a more substantial role in educating their patients.

Keywords Genetic counseling · Preconception counseling · Genetics

Capsule Parous women score higher on a genetics assessment quiz than nulliparous women but do not site their obstetricians as a source of their genetic knowledge.

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Introduction

With the rapid evolution of genetic technologies and expanding options for at-risk couples, genetic counseling and testing are becoming increasingly important aspects of preconception and obstetrical care. Obstetrician/gynecologists (OB/GYNs) have the opportunity to play a pivotal role in providing preconception care for their patients, including evaluating health risks and educating patients about behavioral, environmental, and genetic factors that can contribute to pregnancy outcomes [1]. The American College of Obstetricians and Gynecologists (ACOG) recommends taking a 3-generation family history and asking questions about ethnic background as part of a thorough evaluation of genetic risk [2–6]. Because many pregnancies are unplanned, ACOG also recommends preconception counseling for all women of reproductive age as part of well-woman care [1]. Early risk assessment and patient education regarding genetic risk allow for timely informed decision making. Options for at-risk individuals and couples include adoption, donor gametes, preimplantation genetic diagnosis, avoidance of pregnancy, or prenatal diagnosis.

Few studies assess how well genetic counseling is incorporated into general obstetrics and gynecology. A survey study by Wilkins-Haug et al. in 2000 found that 65 % of OB/GYNs were not confident in their knowledge of genetics, and 40 % considered genetic issues to be the lowest priority in office visits [7]. In a related study, the same authors showed that OB/GYNs have a strong knowledge-base regarding risks of aneuploidy and neural tube defects, but tested poorly on information related to single gene disorders. The physicians cited the rate of change in genetic testing as their biggest obstacle to providing accurate information for their patients [8]. These prior studies all focused on the physicians providing the counseling. The present study focuses on the patient. Specifically, the purpose of this study is to assess genetic knowledge in reproductive-aged women and to determine the role played by their OB/GYNs in their education.

Materials and methods

Subjects were chosen from a group of voluntary survey responders set up through an Internet survey website. Informed consent was obtained from survey respondents. Survey responders revealed demographic information to the survey website and were chosen to fill out surveys according to their demographics. Responders were rewarded with a 50-cent donation to a charity of their choice and a chance to enter a $100 sweepstakes provided by the Internet survey website. Female subjects ages 18–45 were eligible if they fell into one of the following categories: had a child biologically related to them between the ages of 0 and 1 (parous group) or had no children (nulliparous group/reference group). Respondents with the appropriate demographic criteria were emailed a link to the survey by the survey website, and their responses were anonymized. Investigators had no access to subjects' personal information other than their responses to survey questions. To ensure accurate group representation, the first question of the survey asked about parity. Subjects were excluded if they did not meet the defined criteria for either the parous or nulliparous group.

The survey contained 31 multiple-choice questions, addressing the following topics: demographics, the source of subjects' knowledge of genetics, personal experience with genetic testing or disease, and preferences on genetic testing and counseling. Knowledge about genetics was assessed with a 6-question genetics quiz that tested concepts on single-gene disorders and available technologies (see Appendix A). The quiz was divided into 3 fundamental questions and 3 advanced questions. This genetics quiz was validated by 21 general practice obstetrician/gynecologists recruited from 3 academic institutions and 3 community hospitals. OB/GYNs were sent the survey via direct email from the investigators, and their responses were de-identified.

A sample size calculation determined that 178 subjects were needed in each group to demonstrate a 17 % difference among the test groups taking a 6-question quiz (1 question difference). Standard deviation assumption was based on a random number table generated for this test assuming the reference group would score between 3 and 6 correct answers; alpha and beta were set at 5 and 80 %, respectively. The survey was distributed via email in February of 2013 to 450 respondents, 225 parous and 225 nulliparous, to allow for approximately a 10 % dropout rate. Data were analyzed using paired t-tests for continuous variables and chi square for categorical measurements. Confidence intervals of 95 % were used.

Results

There were 450 respondents that began the survey. Of those, 207 responded that they have a child born between January 1, 2012 and January 31, 2013 that is biologically related to them, and were included in the parous group. There were 221 that reported having no children, and were included in the nulliparous group. There were 17 respondents that were classified as nulliparous by the survey website but had a child born after January 31, 2013 and were thus excluded. There were 5 respondents excluded for having a child that was biologically unrelated to them. Of all respondents, 203 parous women completed the survey and were included in analysis (response rate 98 %), and 212 nulliparous women completed the survey and were included in analysis (response rate 96 %).

The age distribution of all respondents was as follows: 16.4 % between ages 18–20, 18.8 % between ages 21–25, 25.8 % between ages 26–30, 21.1 % between ages 31–35, 13.6 % between ages 36–40, and 4.2 % between ages 41–45. The yearly income distribution of respondents included: 22.2 % at less than $20,000, 17 % from $20,000 to $34,999, 18.2 % from $35,000 to $49,999, 17.5 % from $50,000 to $74,999, 6.1 % from $75,000 to $99,000, 5.7 % from $100,000 to $149,000, 3.8 % at $150,000 or more, and 9.7 % chose not to reveal their income. In response to questions regarding demographics, 46 % of respondents reported they were married, and 57 % of respondents reported having a college or graduate degree. With regards to race and political affiliation, 90.5 % of respondents identified themselves as Caucasian, and 32.6 % of respondents described their affiliation as moderate. Amongst those respondents included in the analysis, there were no significant differences between the parous and nulliparous groups in terms of age, race, or income (Table 1). Only 3.8 % of all respondents were pregnant at the time of the survey (5.8 % of parous and 1.8 % of nulliparous women) and 9.2 % of all respondents were actively attempting conception (10.2 % of parous and 8.3 % of nulliparous women). Of all respondents, 50 % were planning on trying to conceive at some point (44.2 % of parous and 55.6 % of nulliparous women).

Table 1 Demographics

	PAROUS n=207 (%)	NULLIPAROUS n=221 (%)	p-value
AGE			
<35	162 (78 %)	182 (82 %)	ns
>35	39 (19 %)	37 (17 %)	ns
RACE			
American Indian, Alaskan native	4 (2 %)	5 (2 %)	ns
Asian	8 (4 %)	21 (10 %)	ns
Black/African American	10 (5 %)	12 (5 %)	ns
Native Hawaiian/Pacific islander	2 (1 %)	6 (3 %)	ns
White	189 (91 %)	194 (88 %)	ns
INCOME			
<$75,000	151 (73 %)	166 (75 %)	ns
>$75,000	45 (22 %)	21 (10 %)	ns

Respondents were permitted to select multiple answers for race

When asked about preconception counseling, 34.5 % of parous women and 11.1 % of nulliparous women recalled counseling by their physicians. Parous women scored significantly higher than nulliparous women on the fundamental genetics quiz (70.9 vs 61.9 %, $p<0.05$). This difference remained but was no longer significant when the 3 advanced questions were included in the scoring (47.7 vs 41.9 %, NS). Parous women scored higher on 5 of the 6 questions.

Parous and nulliparous women were significantly different in citing their physician as a source of genetic information. Among parous women, 38.9 % listed their physician as a source of genetic information compared to 8 % of nulliparous women. Within the parous group, those who listed their physicians as a source of information scored 71.3 % on the 3 fundamental questions, while those that did not list their physician scored 70.1 %. Within the nulliparous group, those that listed their physician as a source scored 68.6 %, those that did not scored 61.4 %.

Of all respondents, 78.1 % stated they would prefer to get genetic information from their physician over other sources. When asked to recall whether they discussed genetic screening with their physicians, 63.1 % of parous and 5.6 % of nulliparous respondents answered affirmatively ($p<0.5$). Of all respondents, 21.4 % noted that they were aware of genetic conditions that run in their family. When asked which type(s) of genetic condition(s), 19 of 84 responses included single gene disorders.

Of all respondents, 44.1 % wished their OB/GYN talked to them more about genetic conditions, and 65.8 % of respondents stated were interested in having carrier screening after reading a short informative paragraph. Few respondents (4.4 % of parous and 5.7 % of nulliparous) were aware of direct-to-consumer genetic testing websites. 2 parous and 3 nulliparous individuals had previously undergone genetic testing through one of these websites.

Of 21 generalist OB/GYNs that received the 6-question genetics quiz, 12 responded and 11 completed the survey. OBGYNs scored an average of 100 % on the 3 fundamental questions and 98.5 % on all 6 questions.

Discussion

A number of studies have looked at the effectiveness of genetic counseling, typically by studying patient recall and understanding following a standardized one-on-one counseling session [9–12]. To our knowledge, the current study is the first to target the general population as an overall measure of counseling effectiveness in practice. This study is also unique in that patient perceptions (not physician perceptions) were examined.

The term preconception counseling implies patient education *prior to* conception. ACOG recommends preconception counseling for all women of reproductive age as part of well-woman care, which should include a discussion about genetic screening [1]. Of the respondents in this study, a small minority of nulliparous women of reproductive age (5.6 %) recalled being offered genetic screening, and only 8.0 % cited their physician as a source of any genetic information. Additionally, only 44.2 % of women who have attempted conception recalled receiving pre-conception counseling at all. In order to deliver adequate pre-conception care, physicians must have sufficient time in the office and patients must be encouraged to seek routine well-woman care outside of pregnancy.

In terms of counseling during pregnancy, this study demonstrates that recently parous women do score higher on a test of genetic knowledge, but they do not credit their physicians a main source of information. Pregnant or parous women may be learning about genetics through self-education and alternative resources such as the Internet. Alternatively, physicians may be appropriately counseling their patients, but patients may not recall the counseling, or do not consider it an important source of their genetic knowledge.

Regardless, the data supports the notion that patients desire to learn about genetics from their physicians. This is in contrast to the patients' perceptions of the care they are receiving, according to the study. OB/GYNs may not be meeting the expectations of not only their patients, but also standards of care.

A potential weakness of our study is that this survey has not been previously validated. In addition, the patient population was primarily white and non-Hispanic, thus limiting the generalizability of these findings to populations that are more racially diverse. The primary strength of this study is that it is the first of its kind to look at genetic knowledge in a general population of reproductive-aged women.

In distributing the genetics quiz to OB/GYN generalists, the goal was to demonstrate that the questions were not so difficult that physicians themselves could not answer them. The sample size was quite small, but we found near perfect scores on the quiz universally between academic and non-academic institutions. Though the quiz was geared more towards the level of a patient, these findings are reassuring in terms the appropriateness of the questions themselves. In addition, the findings demonstrate that the problem may not lie with physician knowledge, but rather that physicians may not be appropriately distributing this knowledge to their patients [7, 8].

As genetic counseling and testing are becoming increasingly important aspects of obstetrical care, the generalist OB/GYN, as a trusted source of genetic information, should play a critical role in educating patients [1, 7, 8]. Future directions should be aimed at examining practices to determine how consistently physicians are offering preconception genetic counseling and making referrals to genetic counselors as appropriate. Barriers to incorporating genetic counseling into routine practice include: rapid changes in available technologies, physician education, and time limitations in the office, and the costs [1, 2, 7, 8, 13].

This study demonstrates need for emphasizing genetic education to physicians as part of their OB/GYN training. In light of advances in pre-implantation genetic testing and increasing promotion of targeted genetic screening in popular media and the internet, a balanced view of this information should be offered by physicians as part of their preconception counseling [14]. This study answers the initial question it poses of "do patients learn about genetics from their obstetrician gynecologists?" The data shows that the majority of subjects do not cite their obstetrician gynecologists as a main source of genetic information.

Compliance with Ethical Standards Conflicts of Interest: John E Buster is the medical director of Previvogententics LLP and a shareholder in that company. Previvogenetics is a medical device company applicable to the field of genetic diagnosis. All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. Informed consent was obtained from all individual participants included in the study.

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APPENDIX A

Quiz Questions

1. If neither you nor your partner has been diagnosed with a genetic disease, is it possible for your children to inherit one?
   a. Yes
   b. No
   c. Don't know
2. How is Cystic Fibrosis most commonly transmitted?
   a. Direct contact with an affected person
   b. Inherited from parents
   c. Environmental exposures
   d. It is not known
   e. Don't know
3. What kind of test is typically used to determine if someone is a carrier for sickle cell anemia?
   a. Blood
   b. Urine
   c. X-ray
   d. Bone marrow biopsy
   e. Don't know
4. Disease X is inherited in an autosomal recessive fashion. If 2 partners are both silent carriers for disease X and they have a child together, what is the likelihood that child will have disease X?
   a. 100%
   b. 50%
   c. 25%
   d. 10%
   e. Don't know
5. To your konwledge, is there a way to avoid pregnancy affected by genetic disease X if both partners are silent carriers and they wish to have a child that is biologically theirs?
   a. Yes
   b. No
   c. Don't know

6. If two partners are both silent carriers of genetic disease X and they wish to avoid a pregnancy affected by the disease, which technology is their best option to have a child that is genetically theirs?

    a. Preimplantation genetic diagnosis (PGD)
    b. Intracytoplasmic sperm injection (ICSI)
    c. Intrauterine Insemination (IUI)
    d. Chorionic Villus Sampling (CVS)
    e. There is currently no way to avoid a pregnancy affected by the disease
    f. Don't know

    Of Note: Questions 1-3 were considered fundamental, 4-6 were considered advanced. Answer choices were randomized for each respondent. Respondents could not move backwa

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