

Specialist physicians' referral behavior regarding preimplantation genetic testing for single-gene disorders: Is there room to grow?

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Objective: To assess whether primary care specialists' demographics, specialty, and knowledge of preimplantation genetic testing for monogenic disorders (PGT-M) influence their practice patterns.

Design: Cross-sectional survey study.

Setting: Academic medical center.

Patient(s): Not applicable.

Intervention(s): None.

Main Outcome Measure(s): Objective PGT-M knowledge, subjective comfort with PGT-related topics, PGT care practices (discussions/referrals), and PGT-M implementation barriers.

Result(s): Our survey had 145 respondents: 65 obstetrician/gynecologists, 36 internists, and 44 pediatricians. Overall, 88% believed that patients at a risk of passing on genetic disorders should be provided PGT-M information. However, few discussed PGT-M with their patients (24%) or referred them for testing (23%). Over half (63%) believed that the lack of physician knowledge was a barrier to PGT use. In terms of subjective comfort with PGT, only 1 in 5 physicians felt familiar enough with the topic to answer patient questions. There were higher odds of discussing (odds ratio, 3.21; 95% confidence interval, 1.75–5.87) or referring for PGT (odds ratio, 2.52; 95% confidence interval, 1.41–4.51) for each additional 0.5 correct answers to PGT knowledge-related questions. The odds of referring patients for PGT-M were the highest among obstetrician/gynecologists compared with those among the internists and pediatricians.

Conclusion(s): Physician specialty and PGT knowledge were associated with PGT-M care delivery practices. Although most specialists believed in equipping at-risk patients with PGT-M information, <1 in 4 discussed or referred patients for PGT. The low levels of PGT-related care among providers may be owed to inadequate knowledge of and comfort with the topic. An opportunity to promote greater understanding of PGT-M among primary care specialists exists and can in turn improve the use of referrals to PGT-M services. (*Fertil Steril Rep*® 2021;2:215–23. ©2021 by American Society for Reproductive Medicine.)

Key Words: Preimplantation genetic testing, monogenic disease, survey, obstetrician/gynecologist, primary care specialist

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Preimplantation genetic testing (PGT) with in vitro fertilization (IVF) offers a way for patients carrying single-gene disorders to screen

their embryos before implantation and decrease the risk of passing on these disorders to their children. Preimplantation genetic testing has become an increas-

ingly common component of IVF cycles in the United States, growing in application from 4% of all IVF cycles in 2011–2012 to a part of >20% of all cycles in 2014–2016 (1, 2). The rise in PGT use is owed predominantly to its use for aneuploidy testing, as opposed to its use for testing for monogenic disorders, i.e., preimplantation genetic testing for monogenic disorders (PGT-M).

Prior research investigating perspectives on PGT-M among carrier couples has demonstrated that anywhere

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from 40% to 80% of patients never heard of or received information on PGT (3–6). However, an interview study showed that when at-risk patients were informed about PGT and referred by their providers for it, they felt a sense “empowerment over the uncertainties of their reproductive futures” (7). This empowerment resulted from the perception that the “ability to select embryos free from genetic conditions alleviates stress” and elimination of “psychophysical trauma during selective pregnancy termination” for couples considering that option (7, 8). Moreover, several patients interviewed were disappointed that their physicians did not inform them about PGT, stating that they felt a sense of “disempowerment” by not knowing that it was available (7). In studies in which reproductive-aged patients carrying monogenic disorders were informed about PGT-M, one third to one half were interested in using it for future pregnancies (4–6, 9–11). For example, in a recent survey of parents of young children with sickle cell disease, 55% were interested in pursuing PGT-M for future pregnancies, citing the main reason for their interest as fear of “financial and emotional burden” of having another child with sickle cell disease (12).

Many physician societies agree with these patient perspectives, stating that PGT-M is acceptable and appropriate for couples at a risk of passing on genetic disorders (13). For example, the American Society for Reproductive Medicine’s Ethics Committee’s opinion on PGT for adult-onset conditions has affirmed that using PGT is “a matter of reproductive liberty” that allows for the “medical good of preventing the transmission of genetic disorders” (14). Belief in the importance of PGT is not just limited to obstetrician/gynecologists (Ob/Gyns). The American Medical Association has also stated that “the use of genetic technology to avoid the birth of a child with a genetic disorder is in accordance with the ethical principles associated with physicians’ therapeutic role” (15).

Given the findings that >50% of carrier couples were not counseled on their option of PGT-M, the findings that up to 50% of these patients would consider using PGT-M for a future pregnancy when informed, and, finally, the physicians’ consensus that PGT is an ethically acceptable reproductive choice to avoid passing on heritable disorders, we wished to investigate the potential reasons underscoring why PGT-M is not used more. To do this, we assessed the perspectives of physicians, who most often act as gatekeepers of patient referrals: specialists providing primary care services. Primary care specialists, specifically internists, pediatricians, and Ob/Gyns, often act as a healthcare “home base” for their patients, interfacing with them multiple times throughout their life and reproductive years. Furthermore, the use of genetic information in primary care is rapidly increasing as direct-to-consumer genetic testing, and with it an increase in the use of preconception-based expanded carrier screening, becomes more widely available (16, 17). With the oncoming swell of this technology, patients are turning to their trusted primary care specialists to interpret carrier screening results, understand inheritance risks, and receive guidance from them on the appropriate reproductive service providers, regardless of their physicians’ prior education on the topic (18, 19). In this capacity, these specialists can be some of the most likely primary care physicians to

encounter patients who might benefit from information on and referrals to PGT-M services.

To date, only one study in the United States has reviewed primary care specialists’ perspectives on the use of PGT-M, specifically among internists (20). In this study, 20%–30% of those surveyed indicated that they referred patients for PGT for monogenic disorders, such as cystic fibrosis or breast cancer type 1 (*BRCA*) mutation. However, only 5% of these internists stated that they had actually suggested PGT to a patient previously. This again highlights the discrepancy in the acceptance of PGT-M versus referrals for the use of PGT-M services. With this in mind, our survey focused on 3 primary care specialist groups (internists, pediatricians, and Ob/Gyns) and aimed to determine if a provider’s demographics, specialty, or prior knowledge of PGT-M influences their discussions with patients and referral practice patterns. A greater understanding of the factors involved in PGT-related care delivery practices among these specialists can help elucidate gaps that need to be filled to maximize appropriate referrals for PGT-M services.

MATERIALS AND METHODS

Study Population and Data Collection

Cross-sectional, descriptive surveys were conducted among 3 specialist groups (internists, pediatricians, and Ob/Gyns) affiliated with the University of Texas Southwestern Medical Center from February to May 2020. Eligible participants included all attending physicians and fellows from the Departments of Internal Medicine, Pediatrics, and Obstetrics and Gynecology. This included subspecialty physicians in each department as well. The surveys were filled out by either respondents attending various departmental grand rounds, where surveys were completed on paper, or those responding to an online REDCap survey, sent out by departmental administrators. The switch from a paper to an online format was because of the coronavirus disease 2019 pandemic. Participation was voluntary, with no reward or incentive offered. Additionally, the survey was anonymous and short, taking an average of 5–10 minutes to complete. The project was reviewed and exempted from approval of the institutional review board at the University of Texas Southwestern Medical Center based on their exemption criteria (STU-2019-1220).

Measures

Based on the survey of these 3 primary care specialists, our study proposed the following questions: first, do physician demographics (i.e., age, institution, and specialty) affect the likelihood that they possess knowledge of PGT? Second, do physician demographics influence the odds of prescribing certain PGT practices and referral behaviors? This question included whether or not demographics influenced which specific PGT indications physicians found acceptable and if they influenced what respondents viewed as barriers to PGT use. Third, we asked if the physicians’ knowledge of PGT influenced the likelihood of prescribing different care delivery practices. For example, did having objective knowledge of or subjective comfort with the topic increase the likelihood

of discussing it with patients and/or referring them for the procedure?

The survey questions are included in the tables for full item review. Specifically, [Table 1](#) details the demographic questions, [Table 2](#) details the PGT knowledge-related questions, and [Supplemental Table 1](#) (available online) details the questions regarding care delivery practices. Briefly, the study variables are described as follows.

Dependent variables. The survey included items to assess 2 distinct domains: (1) physicians' objective and subjective knowledge of PGT and (2) PGT care delivery practices.

Objective PGT knowledge. The participants were asked 4 PGT knowledge-related questions: (1) a true/false question defining PGT, (2) a multiple-choice question on which professional performs PGT (geneticist, embryologist, or fertility specialist), (3) a multiple-choice question on the success rates of pregnancy after PGT (21), and (4) a binary question on whether PGT-M is offered at our academic medical center. For question 2, half a point was given for answering either embryologist or fertility specialist and full point was given for answering both. Correct answers were summed across the domains for a possible continuous score ranging from 0 (0% correct) to 4 (100% correct). Each integer increased by 0.5 points.

Subjective PGT knowledge. The participants were asked whether they felt that they possessed enough basic knowledge to adequately answer patient questions on PGT. The responses were dichotomous: [1] = yes or [0] = no.

PGT care delivery practices. The physicians were asked to report (1) whether they had ever discussed PGT with a patient before and (2) whether they had ever referred a patient for PGT. The responses were dichotomous: [1] = yes or [0] = no.

Independent variables. These included the physicians' age (birth year before 1950, 1951–1970, and after 1970), year of

medical school graduation (before 1980, 1981–2000, and after 2000), practice level (fellow or attending), and practice specialty (Ob/Gyn, pediatrics, or internal medicine). We also included current and previous institutional affiliations (academic, private practice, or both). Regarding this, our academic medical center comprises 2 hospital systems: a public county hospital and a private practice medical campus. Given this, physicians at our institution could be identified as only academicians, only private practice physicians, or a combination of both.

Other descriptive variables. The survey included 3 questions based on the physicians' perspectives: (1) whether they currently treated patients who carried single-gene disorders, followed by whether or not they believed that patients or parents of patients at a risk of passing on a heritable disorder should be provided information on PGT-M; (2) their beliefs on the acceptability of using PGT-M among the following scenarios: avoiding early-life hereditary disorders (i.e., cystic fibrosis or sickle cell disease), avoiding later-life hereditary disorders (i.e., Huntington's disease), avoiding hereditary cancer genes (i.e., *BRCA* or Lynch syndrome), and allowing pregnancy with a savior sibling (one who can provide a bone marrow transplant to a sibling with a fatal disease, such as Fanconi anemia); and (3) opinions on which physician(s) they felt should provide information on PGT to patients. Finally, the physicians were asked to report what they viewed as barriers to PGT use, including the lack of patient or physician knowledge, lack of referrals, lack of patient acceptance, and cost.

Analysis

Univariate statistics (i.e., tabulations and frequencies) were used to describe the independent variables (participant demographics, employment, and training history) and the 2 distinct

TABLE 1

Demographic characteristics of survey respondents across medical specialties (n = 145).

	Medical specialty				X ² or Fisher's exact test
	Ob/Gyn No. (%)	Pediatrician No. (%)	Internist No. (%)	Total No. (%)	
	65 (45)	44 (30)	36 (25)	145 (100)	
Year born					
Before 1950	3 (4)	0 (0)	5 (14)	8 (5)	12.56
1951–1970	22 (34)	16 (36)	15 (42)	53 (37)	
After 1971	40 (62)	28 (64)	16 (44)	84 (58)	
Year of medical school graduation					
Before 1980	6 (9)	1 (2)	7 (20)	14 (10)	8.05
1981–2000	24 (37)	20 (46)	16 (44)	60 (41)	
After 2000	34 (52)	23 (52)	13 (36)	70 (48)	
Practice level					
Fellow	11 (17)	0 (0)	3 (8)	14 (10)	8.71 ^a
Attending	54 (83)	44 (100)	33 (92)	131 (90)	
Institution type					
Academic	43 (66)	30 (68)	21 (58)	94 (65)	4.17
Private practice	1 (2)	3 (7)	0 (0)	4 (2)	
Both	21 (32)	11 (25)	9 (25)	41 (28)	

Note: Ob/Gyn = obstetrician/gynecologist.

^a P < .05.

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TABLE 2

Preimplantation genetic testing knowledge across medical specialties (n = 145).

	Medical specialty			Total No. (%)
	Ob/Gyn No. (%)	Pediatrician No. (%)	Internist No. (%)	
PGT involves in vitro fertilization, biopsy of cells to determine if they carry single-gene mutations, and transfer of unaffected embryos.				
True	62 (95)	41 (93)	36 (100)	139 (96)
False	1 (2)	2 (5)	0 (0)	3 (2)
Which professional performs PGT?				
Embryologist	41 (55)	11 (22)	4 (11)	56 (35)
Geneticist	11 (15)	6 (12)	12 (32)	29 (18)
Fertility specialist	23 (31)	32 (65)	21 (57)	76 (47)
What are the chances of achieving pregnancy after PGT?				
<20%	3 (5)	4 (9)	3 (8)	10 (7)
20%–40%	13 (20)	11 (25)	12 (33)	36 (25)
40%–60%	28 (43)	11 (25)	13 (36)	52 (36)
60%–80%	12 (19)	10 (23)	3 (8)	25 (17)
>80%	8 (12)	6 (14)	5 (14)	19 (13)
Is PGT offered at our medical center?				
Yes	36 (55)	13 (30)	2 (6)	51 (35)
No	2 (3)	1 (2)	1 (3)	4 (3)
Unsure	27 (42)	29 (66)	33 (91)	89 (61)
Objective knowledge: number of correct answers (mean, SE)				
Summation	2.43 (0.09)	1.76 (0.07)	1.97 (0.11)	2.08 (0.06)
Subjective knowledge: do you feel that you have enough basic PGT knowledge to answer patient questions on the topic?				
Yes	21 (32)	7 (16)	1 (3)	29 (20)
No	43 (66)	35 (80)	35 (97)	113 (78)

Note: Ob/Gyn = obstetrician/gynecologist; PGT = preimplantation genetic testing; SE = standard error.

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outcome domains (knowledge and care delivery). The chi-square and Fisher's exact tests were used to evaluate the bivariate relationship among physician demographics, employment history, and medical specialty. Bivariate logistic regression models were used to test the relationship between physician demographics and employment history (independent variables) as well as between physician demographics and discussing and referring a patient for PGT (dependent variables). A bivariate logistic regression model was also used to assess the relationship between medical specialty (independent variable) and subjective knowledge (dependent variable). A bivariate linear regression model was used to analyze the relationship between medical specialty (independent variable) and objective knowledge (dependent variable). Objective knowledge was normally distributed and, thus, treated as a continuous variable with an increasing integer of 0.5 points. When specific survey items were not answered, we excluded those respondents' item from the analyses. All the analyses were conducted using Stata 14.0, and an a priori alpha of 0.05 was used to determine statistical significance (22).

RESULTS

The study population is described in Table 1. We had a total of 145 physician respondents: 65 Ob/Gyns (45%), 36 internists (25%), and 44 pediatricians (30%). Majority of the respondents were born after 1971 (58%), graduated from medical school after the year 2000 (48%), were attending physicians (90%), and were working/had worked in an academic institution (65%).

Supplemental Table 1 displays current care delivery practices, PGT perspectives, and perceived barriers to the use of PGT stratified by medical specialty. In total, 74% of the physicians stated that they treated patients who carried heritable single-gene disorders. Moreover, 88% thought that patients or parents of patients at a risk of passing on genetic disorders should be provided information on PGT. However, only 24% of the physicians surveyed stated that they had ever discussed PGT with their patients, and only 23% had ever referred a patient for PGT. Additionally, PGT-related care practices were infrequent occurrences: majority of the respondents only discussed PGT with their patients (54%) or

referred them to a specialist for the service (80%) once a year. Lastly, in terms of the perceived barriers to PGT use in our study population, over half (63%) felt that the lack of physician knowledge was a major barrier. The other perceived barriers included a lack of patient awareness on the existence of PGT (63%) and its cost-prohibitive nature (75%).

Multiple physician characteristics were found to have an impact on PGT acceptability. Younger physicians (i.e., born after 1970) were significantly more likely to believe that PGT was acceptable to avoid passing on early-life hereditary disorders than their older counterparts ($P < .05$). Physicians who had worked at an academic institution were more likely to believe that PGT is acceptable to avoid passing on early-life hereditary disorders when compared with physicians who had only worked in private practice or both private practice and academic institution ($P < .01$). In terms of the acceptability of PGT to avoid passing on later-life hereditary disorders, this was more likely to be accepted among fellows than among attending physicians and among Ob/Gyns than among internists and pediatricians ($P < .05$).

Physician demographics were additionally seen to affect the perceived barriers to widespread PGT use. Older physicians (i.e., graduated medical school before 1980) were more likely to believe that a major barrier to PGT use was the lack of physician knowledge when compared with younger physicians ($P < .01$). Internists, compared with Ob/Gyns and pediatricians, were also more likely to believe that a major barrier to PGT use was the lack of physician knowledge ($P < .01$). Furthermore, internists were more likely to view the lack of patient awareness on the existence of PGT as a major barrier to its use ($P < .001$).

We subsequently analyzed if physician demographics and knowledge influenced the likelihood of prescribing certain PGT practices and referral behaviors; [Figure 1](#) displays these relationships. First, attending physicians were less likely to discuss PGT with their patients in comparison with fellows (odds ratio [OR], 0.20; 95% confidence interval [CI], 0.06–0.61). Next, medical specialty had a significant effect on the likelihood of discussing and referring for PGT. Compared with the odds of Ob/Gyns discussing PGT with patients, the odds of pediatricians discussing PGT with patients were 0.08 times lower, and the odds of referring patients for PGT were 0.10 times lower. Similarly, the odds of referring were 0.12 times lower among internists than among Ob/Gyns. No other demographic characteristic had a statistically significant impact on referral patterns.

We then evaluated the knowledge of PGT among our study population, stratified by medical specialty, which is seen in [Table 2](#). In terms of objective PGT knowledge, majority of the respondents (96%) knew the correct definition of PGT, and most knew that it was routinely performed by embryologists and fertility specialists (as answered by 35% and 47% of the respondents, respectively). In contrast, only 36% of the physicians correctly answered that the overall success rate of IVF with PGT was 40%–60% (21). Similarly, only 35% of the physicians knew that PGT is offered at our institution. Although over half of the Ob/Gyns (55%) knew that PGT is available at our fertility center, both pediatricians and internists were more often unaware of this service (66% and

91%, respectively), responding that they were “unsure” if it was offered. Among the respondents, the average number of correct answers out of a total of 4 PGT-related questions was only 2.08. Similarly, only 20% of our survey population felt that they had enough basic knowledge of PGT to adequately answer patient questions on the topic.

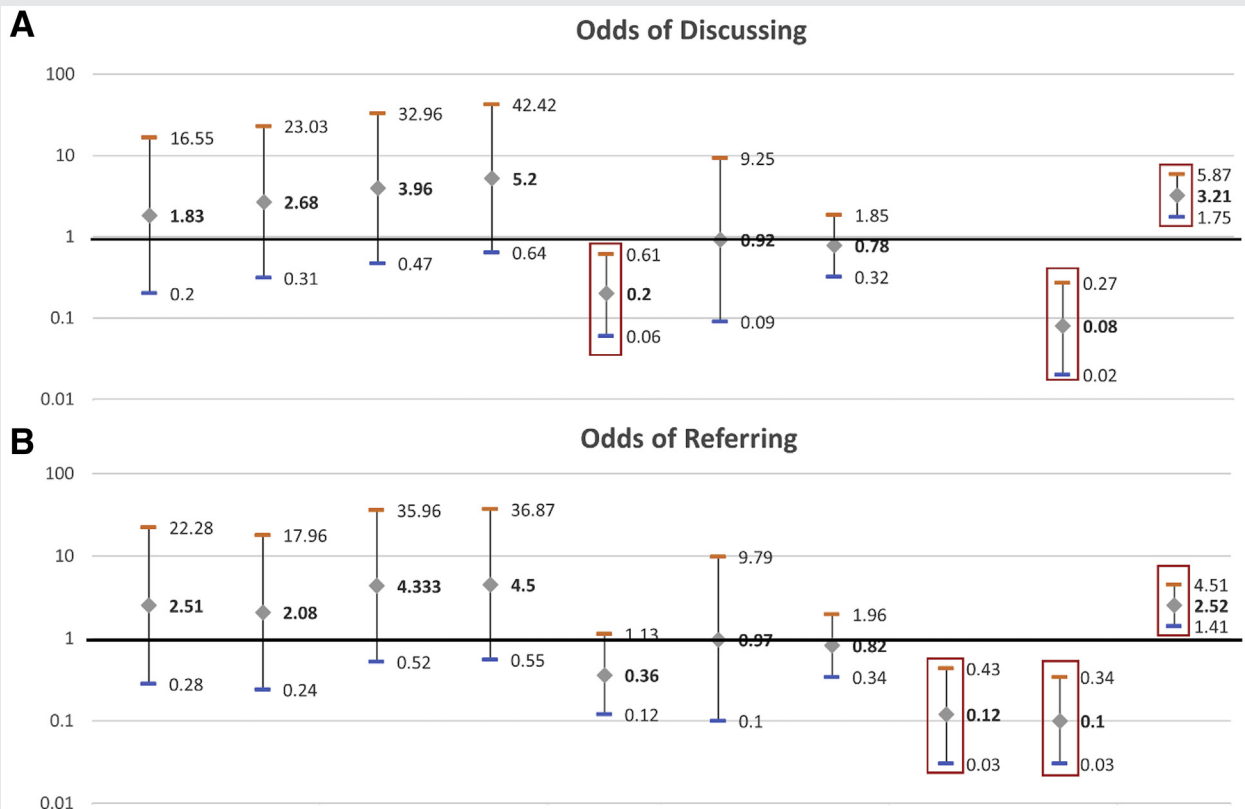
[Table 3](#) displays the odds of PGT knowledge, both objective and subjective, across the medical specialties. Compared with Ob/Gyns, pediatricians were 0.47 times (95% CI, 0.21–0.72) less likely and internists were 0.67 times (95% CI, 0.40–0.93) less likely to answer PGT knowledge-related questions correctly. The odds of having subjective comfort with the topic of PGT were 0.06 times lesser among internists than among Ob/Gyns (95% CI, 0.01–0.46). Although the odds of having subjective knowledge of PGT were significantly lower for internists, they were nonsignificantly lower for pediatricians (OR, 0.41; 95% CI, 0.16–1.07). No other demographic characteristic had an effect on objective PGT knowledge. Lastly, we found that objective knowledge of PGT had a significant effect on practice behavior. Both the odds of discussing PGT with patients (OR, 3.21; 95% CI, 1.75–5.87) and referring patients for PGT (OR, 2.52; 95% CI, 1.41–4.51) were significantly higher for each additional 0.5 correct answers to the PGT knowledge-related questions.

DISCUSSION

Nearly 3 out of 4 primary care specialists in our study treated patients who carried heritable single-gene disorders and nearly 9 out of 10 specialists believed in the importance of providing information on PGT-M to these patients. Despite this, <1 in 4 respondents ever discussed PGT with their patients or referred them for the service. There are many potential reasons why primary care specialists do not discuss or refer patients for PGT-M more frequently. First, several primary care visits are problem-based, and a provider may not feel that it is the appropriate moment or may not have sufficient time to address reproductive counseling in this context. In addition, genetic counseling can be a lengthy discussion, and this may limit specialist engagement. Prior studies on the attitudes of internists toward integrating genetic risk assessments into primary care indicated that “time pressures” and “lack of training” were 2 of the major barriers to integration (23, 24).

With regard to the perceived lack of training, the knowledge-based results of our study raise the concern that specialists’ knowledge of PGT-M might have affected their discussion and referral practices. There are 2 facets to this. First, the specialists had low subjective comfort with the topic, with only 1 in 5 surveyed feeling that they had enough familiarity with PGT to adequately answer patient questions. Indeed, genetic testing for this indication did not gain widespread popularity until the mid-2000s (16). Physicians who completed training before this time might not have been as familiar with its incorporation into practice. On the contrary, younger physicians and fellows, who presumably learned about PGT in medical school and residency, were more apt to believe in its acceptability and, thus, have more discussions with their patients on the matter (16, 24). Second, over 50% of those surveyed believed that the lack of physician knowledge

FIGURE 1



Panel A: Odds of discussing PGT-M with patients
Panel B: Odds of referring patients for PGT-M

Bar 1 ^a	Year born (Ref before 1950)	1951–1970
Bar 2		After 1970
Bar 3	Year of medical school graduation (Ref before 1980)	1980–2000
Bar 4		After 2000
Bar 5 ^b	Practice level (Ref fellow)	Attending
Bar 6	Current and past institutional affiliation (Ref academic)	Private practice
Bar 7		Both
Bar 8	Practice specialty (Ref Ob/Gyn)	Internist
Bar 9		Pediatrician
Bar 10	Number of correct answers to objective knowledge questions	Each additional 0.5 points correct

Odds of discussing or referring a patient for preimplantation genetic testing for monogenic disorders based on physician demographic characteristics, employment history, and objective knowledge of preimplantation genetic testing (n = 145).

Note: PGT-M = preimplantation genetic testing for monogenic disorders; Ob/Gyn = obstetrician/gynecologist.

^aBars are numbered left to right and represent odds ratios and 95% confidence intervals.

^bEncircled bars had statistically significant odds ratios (P < .05).

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was a major barrier to widespread PGT use. In support of this, the odds of discussing PGT with patients or referring them for the service was significantly higher among participants who correctly answered increasingly more knowledge questions. Additionally, our study found that Ob/Gyns, who train with reproductive endocrinologists and genetic counselors during their residency, were more likely to possess more objective

PGT knowledge; express more comfort with the topic; and have more discussions with their patients, leading to referrals, than other primary care specialists.

In our study, 3 out of every 4 specialists reported having treated patients who carried heritable single-gene disorders. Although not chiefly responsible for reproductive counseling, especially during problem-based visits, it might be beneficial

TABLE 3

Preimplantation genetic testing knowledge across medical specialty.

Medical specialty	Objective knowledge OR (95% CI)	Subjective knowledge OR (95% CI)
Obstetrician/gynecologist	Ref	Ref
Pediatrician	0.47 ^b (0.21–0.72)	0.41 (0.16–1.07)
Internist	0.67 ^b (0.40–0.93)	0.06 ^a (0.01–0.46)

Note: CI = confidence interval; OR = odds ratio.

^a $P < .01$.

^b $P < .001$.

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for all primary care specialists to have basic understanding of PGT-M and the appropriate referral services because this group clearly treats a large proportion of patients at a risk of passing on monogenic disorders. Prior clinical reviews on genetic screening in primary care agree with this notion, indicating that these physicians “have an important role to play in helping their patients navigate the rapidly changing terrain of genetic screening services” (25). Specifically, in the preconception population, there has been increasing ease of accessibility to expanded carrier screening, even in couples who have no personal or family history of a disease (26). In a recent systematic review, it was noted that anywhere from 10% to 30% of a general preconception population can be interested in expanded carrier screening (27). Given this, it is critical to improve education for primary care providers on how to discuss next steps with a patient after a positive carrier screen, which includes counseling on the use of PGT-M to avoid passing on heritable disorders. This is especially true as genetics becomes more incorporated into overall health maintenance in a move toward personalized medicine (19).

An additional benefit of expanding the use of PGT could be, as stated based on the American Society for Reproductive Medicine Committee's opinion, the “potential societal benefits of reducing the overall burden of disease” (14). Specifically, the societal benefits of expanded adoption of PGT-M would include not only decreased disease incidence but also national healthcare savings. As an example, one study highlighted possible healthcare savings by performing a cost-benefit analysis on the implementation of a national PGT program to decrease the incidence of cystic fibrosis (28). They compared the one-time average amount that would be spent to deliver an unaffected child via PGT-M for a carrier couple with the lifetime cost of the treatment of a patient with cystic fibrosis. They modeled an overall average savings of \$2.3 million per patient and \$2.2 billion annually for all new patients born with cystic fibrosis when compared with the costs of using PGT-M for carrier couples. They also estimated that over a 37-year period (the average life expectancy of a person with cystic fibrosis at the time of the study), the use of PGT-M could lead to the avoidance of >30,000 new cases of cystic fibrosis and >600,000 “cumulative years of patients suffering” (28). This study showed the possible benefits of broadening the implementation of PGT-M for couples who carry or are affected by one of the 7,000 known monogenic disorders: PGT-M

allows the birth of an unaffected child, decreasing the number of years of medical hardship, and reducing healthcare cost not only for the patient and their family but also on a national level (29). Given these potential patient and societal benefits, one can see the importance of having specialist physicians provide information on this option to interested couples. Of course, all physicians should keep in mind that the decision to pursue PGT-M is a challenging and individualized one and that couples should be supported whether or not they are interested in or deny this option.

Our study has multiple strengths that bolster its clinical applicability in terms of understanding the gaps that need to be filled to expand primary care specialists' comprehension of and referrals for PGT-M. First and foremost, we targeted our survey at 3 primary care providers who often treat patients with monogenic disorders and, thus, might have an important role to play in furthering PGT-M use. To date, there have only been a small number of studies that delved into the issue of primary care specialist knowledge and referral behaviors in terms of PGT. Majority of these studies focused specifically on hereditary cancer genes alone, such as *BRCA* and Lynch syndrome (30–33). Two studies in the United States reviewed the perspectives and referral practices for PGT-M among specialty and subspecialty physicians, specifically surveying internists, psychiatrists, and neurologists (20, 34). In these studies, like our own, there was a discrepancy between the physicians' perspectives on the clinical use of PGT for genetic testing (30%–40% stated that they would theoretically refer their patient for the procedure) and actual discussions occurring in the office (only 3%–5% had ever discussed PGT with a patient).

Our study was able to expand on this previous research of specialists practice patterns by including those who had not yet been studied in the United States but have a place on the frontlines of referrals: pediatricians and Ob/Gyns. One prior study focused solely on PGT perspectives among women's health providers, which included Ob/Gyn physicians, as well as nurses, mid-level providers, and genetic counselors (35). In this study, 94% of the physicians responded that patients should be given the option to use PGT-M to avoid childhood-onset diseases, if interested. This survey also asked the respondents if they had prior knowledge of PGT, with 77% affirming that they did. However, this study did not specify how many of those with prior knowledge were

physicians and did not ask any questions related to objective knowledge of PGT, making it difficult to interpret if their knowledge on the topic was related to their perspectives on the practice. By including questions on both objective and subjective knowledge of PGT, our survey was able to glean a stronger understanding of why there is such a vast difference between acceptance of PGT by specialists and actual referrals by this group for the procedure: an overall lack of comfort with the topic.

There are several ways our study could be built upon to further understanding on primary care specialists' perspectives of PGT-M. Future surveys can be expanded to include more physicians in private practice because it is possible that their practice patterns differ from those of academicians. Other future surveys can also expand beyond the primary care scope and include specialty-specific physicians, such as hematologists, neurologists, and pulmonologists, who likewise develop long-term relationships with patients who carry monogenic disorders. Taking all the cited barriers to widening PGT use into consideration, the next steps forward to bolster primary care specialists' knowledge of and subsequent referrals for PGT-M will likely be a requisite for the efforts of younger physicians to bolster the knowledge of their colleagues through educational efforts.

In conclusion, our study showed that there is an opportunity to promote greater understanding of PGT among all primary care specialists and, with this, increase the number of referrals and the use of PGT-M for patients at a risk of passing on monogenic disorders. With greater comprehension of PGT, primary care specialists can plan the time of annual visits to ask reproductive-aged patients if they are interested in becoming pregnant, either in person or through previsit pamphlets and checklists. If so, the provider can then use preconception carrier screening as a part of their referral algorithm to identify patients who may benefit from PGT-M. Based on our results, it is critical to gain the support of younger Ob/Gyns in practice, who have the most familiarity and comfort with the topic. This group can hopefully set a foundation to strengthen PGT-M care delivery practices among not only other obstetric providers but also their colleagues in internal medicine and pediatrics departments. Using this group, follow-up interventions to advance primary care specialist knowledge of PGT-M can take the form of grand rounds lectures and continuing medical education courses led by both Ob/Gyns and reproductive endocrinologists. If we can take steps to further the awareness and basic comprehension of PGT-M among physicians treating patients with single-gene disorders, we can likely maximize appropriate referrals for PGT-M services for interested patients.

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