Genetics educational needs in China: physicians' experience and knowledge of genetic testing

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Purpose: The aims of this study were to explore the relationship between physicians' knowledge and utilization of genetic testing and to explore genetics educational needs in China.

Methods: An anonymous survey about experience, attitudes, and knowledge of genetic testing was conducted among physicians affiliated with Peking Union Medical College Hospital during their annual health evaluation. A personal genetics knowledge score was developed and predictors of personal genetics knowledge score were evaluated.

Results: Sixty-four physicians (33% male) completed the survey. Fifty-eight percent of them had used genetic testing in their clinical practice. Using a 4-point scale, mean knowledge scores of six common genetic testing techniques ranged from 1.7 ± 0.9 to 2.4 ± 1.0 , and the average personal genetics knowledge score was 2.1 ± 0.8 . In regression

analysis, significant predictors of higher personal genetics knowledge score were ordering of genetic testing, utilization of pedigrees, higher medical degree, and recent genetics training ($P < 0.05$). Sixty-six percent of physicians indicated a desire for specialized genetic services, and 84% reported a desire for additional genetics education.

Conclusion: This study demonstrated a sizable gap between Chinese physicians' knowledge and utilization of genetic testing. Participants had high self-perceived genetics educational needs. Development of genetics educational platforms is both warranted and desired in China.

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Key Words: attitudes; education; experience; genetic testing; knowledge

In China, clinical genetics was developed as part of the family planning system for "improving birth outcome and child development" and is available only in metropolitan hospitals.¹ Genetic testing (GT) is ordered mainly by pediatricians or obstetricians for prenatal or birth defect diagnoses. Recently, the debates about noninvasive prenatal testing for Down syndrome offered by Beijing Genomics Institute have increased the visibility of China and stimulated the attention of the public and the government. In February 2014, the National Health and Family Planning Commission issued a ban on all genetic sequencing services in clinical practice and required sequencing software and equipment to be registered and regulated under China's Food and Drug Administration.² Without clinical utilization standards, GT in China resides in a legal gray zone. At present, GT can only be performed in academic institutions as research tests or in private commercial companies as a direct-to-consumer service for nonclinical use.³

Chinese medical education has three separate training tracks: Medical Doctor, Medical Master, and Medical Bachelor. Physicians can complete specialty and subspecialty clinical training after graduation from any of these programs. The distribution of physicians with these three degrees is variable across different levels of hospitals (primary, secondary, and tertiary) and across provinces. Genetics education in China began in the 1980s when selected medical schools began offering genetics

courses that focused predominately on molecular genetics and had limited clinical content. Currently, there is great variability in the duration and content of genetics curricula among medical schools and across the three training tracks. As a result, some clinicians have never had any genetics education while in medical school. Although the Chinese health-care system is highly subspecialized in other fields, there is no specialty or subspecialty training in clinical genetics. In 2013, the College of Life Sciences at Fudan University established the first genetic counseling pilot training program in China with the goal of developing a master degree program.⁴ At present, there are no graduates of this program practicing as specialty-trained geneticists or genetic counselors.

The explosive advances in genetics mean that GT is now available for both rare and common conditions at many life stages.⁵ The growth in the number of commercial GT companies in China and the advertisements of these services are increasingly forcing physicians in China's health-care system to integrate genetics into general clinical practice.⁶ The fact that there are no specialty-trained health-care providers to provide assistance compounds this challenging expansion. Studies outside of China have shown that physicians' personal knowledge strongly influences their genetics practice.^{7,8} However, no study about Chinese physicians' knowledge of genetics has been reported previously. We designed this survey to begin exploring

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how physicians in China are dealing with the genetics explosion. To this end, we looked at knowledge about and utilization of GT and the genetics educational needs in Peking Union Medical College Hospital, a renowned general hospital founded in 1921 by the Rockefeller Foundation that is considered to be the hospital most closely aligned with modern Western medicine in China.

MATERIALS AND METHODS

Questionnaire design

A three-section questionnaire containing 20 items was designed to assess experience with, attitudes toward, and knowledge of GT; content was derived from review of multiple surveys exploring genetics knowledge in physicians outside of China and with input from US genetics experts. Demographic information (gender, age, years in workforce, highest medical degree, time of last genetics training, and specialty) was also gathered. In the experience section, multiple-choice questions with single answers were used to assess the frequency of collecting family histories (always, often, occasionally, or never), drawing pedigrees (always, often, occasionally, or never), and ordering GT (ever/never). Participants who had ever ordered GT were asked to name the testing methodologies that were utilized. A question with more than one answer was used to explore rationales for testing (i.e., diagnosis, treatment, risk evaluation, and prenatal diagnosis). The attitudes section assessed self-perceived genetics educational needs (yes/no), preferences for localization of specialized genetics services (i.e., "In your opinion, GT should better be performed in: generalized clinics, special genetics services, or either of the above"), and the most frequent barriers to ordering GT in clinical practice (i.e., quality of the technique, knowledge limitation, government medical policies, social or ethical/religious concerns, or there is no need for GT in my clinical practice). Each participant was asked to self-rate his or her knowledge about six categories of GT techniques (karyotypes, fluorescence in situ hybridization/polymerase chain reaction/Southern blot, mutation testing, Sanger sequencing, next-generation sequencing, single-nucleotide polymorphism microarray) using a 4-point knowledge scale, with 1 indicating "no knowledge," 2 indicating "limited knowledge," 3 indicating "greater than average knowledge," and 4 indicating "highly knowledgeable." For each responder, a personal genetics knowledge score (PGKS) was calculated by averaging responses to the six GT knowledge questions. The survey was developed in English and translated into Chinese by one of the authors, a native Chinese-speaking physician.

Participants and recruitment

Subjects were recruited from the 1,300 physicians affiliated with Peking Union Medical College Hospital during their annual health evaluation in the Department of Health Management from 20 March 2014 to 20 May 2014. Health evaluations can occur at any time during the year; ~70 physicians were scheduled for evaluations during the survey time. An associate worker with no medical background was responsible for

study recruitment and questionnaire collection to help minimize discomfort in answering the knowledge questions. All potential participants were approached about the study at the front desk as they checked out and were invited to participate autonomously. The results were translated back into English by the lead author under the supervision of a genetics expert in the University of Michigan Health System.

This study was reviewed and approved by both the Ethic Committee of Peking Union Medical College Hospital (S-673) and the Committee of Human Social and Behavioral Science of the University of Michigan (HUM00087668).

Statistics

Statistical analyses were performed using SPSS version 17.0 (SPSS, Chicago, IL). Descriptive statistical analysis was conducted for all closed-ended questions. Age, knowledge scores for individual GT, and PGKS are presented as means $\pm$ SDs. To support analysis of associations with PGKS, responses to multiple-choice questions with four answers were collapsed into two components. The Mann-Whitney *U*-test was used to identify associations with PGKS in two-component categories, and Kruskal-Wallis analysis of variance was used for analysis of three-component responses. Stepwise multivariate linear regression was performed to identify predictors of higher PGKS. A two-tailed *P* value of <0.05 was considered statistically significant.

RESULTS

Demographics

A total of 66 physicians participated in the survey; 64 completed the entire questionnaire. Demographic characteristics are presented in [Table 1](#).

Experience and attitudes

In response to the question "How often do you take a family history in your clinical practice?" 72% responded "always" ($n = 22$) or "often" ($n = 24$). Pedigree drawing occurred less

Table 1 Demographics

Demographic characteristic	Response	n (%)
Sex	Male	21 (33)
	Female	43 (67)
Age ^a (years)	40 $\pm$ 13	— (—)
Years in workforce	≤ 10 years	27 (42)
	> 10 years	37 (58)
Medical degree	Medical doctor	32 (50)
	Medical master	18 (28)
	Medical bachelor	14 (22)
Last genetics training	≤ 2 years ago	23 (36)
	> 2 years ago	41 (64)
Specialty	Gynecologist/obstetrician	4 (6)
	Pediatrician	2 (3)
	Other	58 (91)

^aOnly 43 of 64 physicians provided their exact age, and age is presented as mean $\pm$ SD.

commonly; 88% responded “never” ($n = 23$) or “occasionally” ($n = 33$). Fifty-eight percent ($n = 37$) of physicians had ordered GT in their clinical practice for “help with diagnosis” (51%), “treatment” (23%), “risk evaluation” (17%), or “prenatal diagnosis” (9%). Knowledge about the actual techniques associated with this testing was variable: 22 of the 37 physicians who had ordered testing were able to name the techniques, 8 of them stated that they did not know what technique was used, and 7 did not answer the question. Sixty-six percent of participants ($n = 42$) expressed the opinion that it would be better to perform GT in specialized genetics settings. Eighty-four percent ($n = 54$) of physicians acknowledged self-perceived educational needs in genetics. The barriers to ordering GT that were most frequently encountered were “knowledge limitation” (31%), “quality of the technique” (25%), “government medical policies” (21%), “social or ethical/religious concerns” (15%), and “no need for GT in clinical practice” (8%).

Knowledge

Participants were asked to rate their knowledge of six common GT techniques. On a scale of 1–4, in which 4 is highly knowledgeable, the mean knowledge scores for each technique were: (i) karyotype, 2.3 ± 1.0 ; (ii) fluorescence in situ hybridization/polymerase chain reaction/Southern blot, 2.3 ± 1.0 ; (iii) mutation testing, 2.4 ± 1.0 ; (iv) Sanger sequencing, 1.7 ± 0.9 ; (v) next-generation sequencing, 1.8 ± 0.9 ; and (vi) single-nucleotide polymorphism microarray, 1.8 ± 0.9 . More than 80% of the participants had “no knowledge” or “limited knowledge” of Sanger sequencing, next-generation sequencing, and single-nucleotide polymorphism microarray. The PGKS of all physicians was 2.1 ± 0.8 . Analysis of factors associated with higher PGKS is presented in **Table 2**. Significant associations were found in participants with more recent entry into the medical workforce ($P = 0.047$), higher medical degrees ($P = 0.001$), regular family history taking ($P = 0.008$), pedigree drawing ($P < 0.001$), GT ordering ($P < 0.001$), and awareness of a need for genetics education ($P = 0.026$). After the multivariate regression, the only factors with significant associations with higher PGKS were GT ordering ($P < 0.001$), frequent pedigree drawing ($P = 0.004$), higher medical degree ($P = 0.024$), and recent genetics training ($P = 0.035$).

DISCUSSION

This is the first study to explore Chinese physicians’ knowledge of GT in their clinical practice. Seventy-two percent of physicians reported they collected family history “often” or “always.” Family history collection is a basic tool in clinical genetics; however, the gold standard is collecting adequate amounts of information and comprehensive interpretation.⁹ Because 88% of participants had limited experience drawing pedigrees, the quality of the family history information may not be equivalent to the collection frequency. Similar results of an imbalance between the frequency and quality of family history collecting were also reported in previous surveys among US physicians.^{10,11} Our finding of an association between higher PGKS

Table 2 Personal genetics knowledge score (PGKS) comparisons

Category	Response	n	PGKS ^a	P value
Sex	Male	21	2.0 ± 0.8	0.370
	Female	43	2.1 ± 0.8	
Years in workforce	≤10 years	27	2.3 ± 0.7	0.047
	>10 years	37	1.9 ± 0.8	
Medical degree	Medical doctor	32	2.4 ± 0.7	0.001
	Medical master	18	1.8 ± 0.7	
	Medical bachelor	14	1.6 ± 0.6	
Last genetics training	≤2 years ago	23	2.2 ± 0.8	0.473
	>2 years ago	41	2.0 ± 0.8	
Family history collection	Always/often	46	2.3 ± 0.8	0.008
	Occasionally/never	18	1.7 ± 0.6	
Pedigree drawing	Always/often	8	3.1 ± 0.4	<0.001
	Occasionally/never	56	1.9 ± 0.7	
GT ordering	Ever	37	2.5 ± 0.7	<0.001
	Never	27	1.5 ± 0.5	
Self-perceived educational needs	Yes	54	2.2 ± 0.7	0.026
	No	10	1.7 ± 0.9	

GT, genetic testing.

^aPGKS is presented as mean $\pm$ SD.

and GT ordering is consistent with those of previous studies.¹² Physicians with more genetics knowledge are more confident integrating new GT techniques into their practice. Increased exposure to genetic conditions can also encourage physicians to seek more education and result in higher knowledge levels.

Developments in molecular genetics and implementation into clinical practice are occurring rapidly and creating pressure on non-genetics trained clinicians.¹³ Self-perceived genetics knowledge deficits by general practitioners are a global problem.^{14,15} We might suppose that educational needs would likely be addressed in those with lower knowledge levels; however, the finding that the group of physicians with self-perceived educational needs had relatively higher PGKS suggests that the gap between knowledge and practice may be larger than expected. Even in developed countries, current medical school courses may be insufficient to ensure competence in the practice of genetics.¹⁶ The finding that 84% of physicians in this study identified a need for genetics education suggests that current medical genetics education in China is inadequate; future studies need to determine how to provide this education to medical trainees and practicing physicians.

The prevalence and spectrum of genetic diseases in China are similar to those in Western populations.^{17,18} With the Chinese population’s positive attitudes toward GT¹⁹ and increasing willingness to pay for health care, there is little doubt that genetics will be infused into nearly every aspect of health care in China, similar to the current situation in Western countries. Our results demonstrate that without help from specialty-trained clinical geneticists or genetic counselors, general physicians in China who are practicing clinical genetics have insufficient knowledge and confidence. The physicians who participated in

this study seem to already understand this challenge. Most participants thought that GT should be performed by specialized genetics services and desired additional education. Successful development of genetics training programs in other parts of the world has demonstrated the importance of developing genetics-focused education in response to local factors.²⁰ Our findings suggest that further research regarding Chinese genetics educational needs is required and that the Chinese health-care system may need to place greater emphasis on developing genetics educational platforms in the future.

Limitation

The major limitation to this study is the possible restriction on the generalization of results to the whole health-care system in China because this survey was performed among only 66 physicians in one hospital.

DISCLOSURE

The authors declare no conflict of interest.

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