The rise of the genetic counseling profession in China

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1 INTRODUCTION

The development of the genetic counseling profession is at different stages across the world (Ormond et al., 2018). Since the geneticist Sheldon C. Reed first coined the term “genetic counseling” in 1947 (Reed, 1974), the United States has been the leading pioneer in the field of clinical genetics, and has set an excellent model for the development of the genetic counseling profession. The United Kingdom established its own professional association: the Association of Genetic Nurses and Counselors at the end of the 1980s, and this association now has more than 300 well-trained and registered genetic counselors (Skirton et al., 2013). However, most other European countries lacked an accreditation system for genetic counseling and there were no genetic counselors in Germany, Hungary, Turkey or Czech Republic as recently as 2012 (Cordier, Lambert, Voelckel, Hostery-Ugander, & Skirton, 2012).

In Asia, Japan founded the Japanese Society of Genetic Counseling in 2001 and currently has more than 100 genetic counselors (http://www.jsgc.jp). The genetic counseling profession in other Asian countries, such as Korea, Singapore, Thailand and Malaysia, is still in its infant stages (Laurino et al., 2018). While the development of the genetic counseling profession in China is also very preliminary, recent strong support from the government and concerted efforts of overseas experts and domestic geneticists and clinicians have inspired breakthroughs in the field of clinical genetics over the last two to three years.
In this report, we first provide a brief review of the history of Chinese medical genetics and then describe recent opportunities, endeavors, and achievements. The current challenges and reflections upon future development are also discussed.

2 | MAIN TEXT

2.1 | Brief history of medical genetics in China

Genetic counseling is a process of helping people understand and adapt to the medical, psychological, and familial implications of genetic contributions to diseases (National Society of Genetic Counselors’ Definition Task et al., 2006). Medical genetics is a branch of medicine that involves the diagnosis, management, and counseling of patients with genetic disorders. Therefore, as an essential part of medical genetics, the development of genetic counseling relies much on that of medical genetics. We briefly review China’s development of medical genetics here in order to offer the context of the development of genetic counseling in China.

As early as 1937, Chinese scientists had already begun to study genetics by analyzing the frequency of the color blindness gene in the Chinese population (S. Z. Huang & Gao, 2006). Before 1960, most clinical reports were case studies involving a few genetic diseases, such as G6PD deficiency. In 1962, the medical geneticist Min Wu established the division of Human Cytogenetics at the Department of Pathology in the Institute of Experimental Medicine, Chinese Academy of Medical Sciences, meanwhile, Dr. Xiao-qian Zhang and Dr. Hui-yuan Luo established the Division of Medical Genetics at the Internal Medicine Department in Peking Union Medical College Hospital. Subsequently, more and more domestic doctors and geneticists became involved, including Dr. Zu-fei Su, who investigated Down syndrome (Su et al., 1963), Dr. Yi-tao Zeng, who investigated abnormal hemoglobin diseases (Zeng, 2002) and Dr. An-guo Han, who performed chorionic villus sampling (Sun, Wang, Wu, & Ning, 1985). Dr. Jia-hui Xia opened the first clinic to offer genetic counseling services at Xiangya Hospital in Central South University in 1972.

In 1978, the Chinese government implemented the Reform and Opening-up policy encouraging further development of medical genetics. This led to the founding of the National Committee of Human and Medical Genetics under the leadership of the Chinese Society of Genetics in 1979. This committee consisted of eight specialty groups among which groups of internal genetic medicine, pediatric genetic medicine, neuropsychiatric genetic medicine and prenatal diagnostic medicine were closely related to clinical genetics (C. G. Li, 2004). Seven years later, the medical section of the committee formed an independent society, the Association of Medical Genetics, under the leadership of the Chinese Medical Association (C. G. Li, 2004). During this period, several medical colleges, including Sun Yat-sen Medical University, opened new departments to teach medical genetics. Nationwide surveys of congenital anomalies and genetic diseases were also actively conducted (S. Z. Huang & Gao, 2006). In the early 1980s, more clinics opened offering genetic counseling services. These clinics were mainly staffed by pediatricians, obstetricians and other healthcare workers, who were briefly trained in medical genetics. Karyotyping was the main genetic testing performed. Approximately 1,200 abnormal karyotypes were collected in China in these clinics between 1985 and 1990 (P. Li, 2006). In 1987, the Polymerase Chain Reaction (PCR) method was introduced in China which greatly facilitated the development of clinical molecular genetics. More and more genetic disorders, such as Duchenne muscular dystrophy and Huntington disease, began to be tested and diagnosed in clinics.

As for governmental regulation, it is not until 2002 the Ministry of Health of the P.R. China issued a document titled “The Management Means for Prenatal Diagnosis” which was the first governing document specified for clinical genetics and relevant techniques. It ensured the statutory application of clinical genetic tests and genetic counseling in prenatal diagnosis. After this milestone event, advancements of the clinical applications of medical genetics began to slow, while laboratory technologies and scientific research on medical genetics began to rapidly advance.

In 1998, Prof. Jia-hui Xia and his team cloned the GJB3 gene and associated this gene with bilateral high-frequency hearing impairment. This work was published in Nature Genetics and described the first human pathogenic gene discovered and cloned in the Chinese population (Xia et al., 1998). During this period, a group of geneticists working abroad returned to China further enhancing the genetic research in China. In 2001, Prof. Lin He and his team linked the first human Mendelian trait, that is, human brachydactyly to the IHH gene in Chinese individuals (Gao et al., 2001). Additionally, his team found a novel gene locus responsible for the agenesis of permanent teeth (He-Zhao deficiency) representing the first genetic disease named after Chinese (Liu et al., 2001). Since these discoveries, more genes have been cloned, and their pathological functions scrutinized in the context of various genetic diseases in the Chinese population. This research boom in medical genetics was further encouraged by China’s involvement and accomplishment in the Human Genome Project (Cynaroski, 2001). Subsequently, increasing numbers of research papers on various genetic diseases were successively published by Chinese researchers in world-class top-tier journals such as Nature, Science, and NEJM (Chen et al., 2003; Gao et al., 2009; H. L. Huang et al., 2014).

2.2 | Genetic counseling in China around 2015 and new opportunities

Although genetic research rapidly developed in China in the first decade of the 21st century, the translation of these research findings to the clinics remained static. There was a disconnection between research and clinical application of medical genetic technologies, also a lack of understanding of the importance of genetics in modern medicine by the public and the government. Consequently, few students choose to take this profession, there was a lack of vision and guidance for the development of clinical genetics or genetic counseling in China.

At the end of 2013, the State Council finally released a document titled “Guidelines on the establishment of a standardized training system for resident” opening a door for medical students with training in medical genetics to perform standardized clinical services in hospitals (http://www.nhfpc.gov.cn). In 2014, the National Health and Family Planning Commission further recognized medical genetics as the 27th secondary clinical specialty and requested that major maternal and child
health care hospitals establish clinical departments specifically dedicated to medical genetics (http://www.moh.gov.cn). Moreover, the Chinese Medical Geneticists Association's (CMGA) annual meeting in 2018 initiated a Medical Education Post-Graduation Program especially designed for six classes of genetic diseases. These systematic changes for training clinical geneticists are greatly boosting the number of domestic clinical geneticists, which has set a good precedent and is paving a solid way for professionalization of genetic counseling in China.

Genetic counselor is not yet a formal profession in China as of today. Genetic counseling services which are experiencing a rapid increase of demand, are scarcely available only in a few large cities such as Shanghai (http://sjff.stats-sh.gov.cn/zwy/rpt.html). And even in Shanghai, according to a recent survey conducted by Shanghai Center for Women and Children's Health, there were only 52 quasi genetic counselors providing services on traditional genetic diseases (such as Down Syndrome) by regular genetic testing (such as karyotype analysis). Most of them were at their middle age, majoring in nongenetic medical specialties with no systematic training background of modern genetics or even genetics. Due to the large gap in the development of the genetic counseling profession between China and developed countries, several overseas medical geneticists and genetic counselors have exerted great effort to introduce clinical genetics to their colleagues in China. The Association of Chinese Geneticists in America has played a leading role in promoting the status of medical genetics and educating a new generation of medical geneticists by directly introducing the principles and practices of medical genetics and genetic counseling in the United States to China. Other organizations, such as the American College of Medical Genetics (ACMG), have also aided in the process. Domestic geneticists driven by a strong sense of social responsibility have spared no effort in facilitating this progress. As a member of the National Committee of the Chinese People's Political Consultative Conference, Prof. Lin He proposed and appealed repeatedly in conferences to implement the clinical applications of genetic tests and genetic counseling to prevent congenital anomalies in China.

These concerted efforts and the gradual maturation of the domestic medical genetic environment have finally catalyzed a number of great turning points in 2015. In January 2015, nearly one year after the ban by Chinese authorities forbidding any clinical application of Next Generation Sequencing (NGS) techniques, the National Health Commission eventually offered a list of 109 pilot institutes that could conduct noninvasive prenatal tests (NIPT) for prenatal genetic screening (http://www.nhfpc.gov.cn). On February 9, 2015, the Chinese Board of Genetic Counseling (CBGC) was founded (http://www.cbgc.org.cn).

Just two months later, the CBGC launched its first training course in Shanghai introducing basic genetic counseling concepts. This course perfectly aligned with the expansion of NIPT at hospitals and attracted hundreds of hospital directors and medical doctors. To meet such demand across the country, the CBGC held three more sessions in different provinces.

In addition, on October 29, 2015, the Central Committee of the Communist Party of China announced the unbidden "universal two-child" policy, which ended the "one-child" policy that began in 1980. This important policy shift is expected to lead to an increase in the number of newborns in China (http://www.chinanews.com/gn/2015/10-29/7596619.shtml). The advanced maternal and paternal ages of those who are having the second child imposed a significantly increased risk of birth defects and severe genetic disorders. As a result, the demand for genetic counseling becomes overwhelming. Moreover, the establishment of the CMGA at the end of 2015 consolidated the professional status of medical geneticists. We hope this will in turn further facilitate the eventual professionalization of genetic counseling in China.

### 2.3 Objectives and activities of the CBGC

The aims of the CBGC are to set up standardized workflows for genetic counseling, train eligible genetic counselors and enhance the standardization, professionalization and normalization of genetic counseling in China. Ultimately, well-trained genetic counselors and qualified genetic counseling programs will enable patients and family members to make informed decision which in turn will lead to the reduction of birth prevalence of severe congenital anomalies and genetic disorders. To better achieve these aims, the CBGC chose a pragmatic strategy to meet the most urgent needs in genetic counseling across the country and to simultaneously introduce advanced materials and experiences from other countries to the domestic health system and public (Figure 1).

We designed short-term courses to meet the demand of training a large number of practitioners in the field of NIPT which were permitted to expand by the government in 2017. These courses invited well-known medical genetics experts at home and abroad to introduce the basic concepts of genetic diseases, key processes in genetic tests, general rules and skills in genetic counseling and common applications of medical genetics in various diseases. Unsurprisingly, the courses were warmly received. Until June in 2018, 16 sessions of basic and intermediate-level courses have been carried out in cities in 13 provinces across China (Figure 2). Meantime, the CBGC encouraged top hospitals to become accredited training institutions for genetic counselors. Until June in 2018, 24 hospitals have been authorized as the training bases. Moreover, the first large-scale comprehensive demonstration base for genetic counseling in China was established at the Shanghai Center for Women and Children's Health with the CBGC as the consultative committee. In October 2016, this base organized the first worldwide remote genetic counseling event featuring tele-consultations and providing world-class genetic counseling services to local patients in a safe and cost-effective manner. A more comprehensive network interconnecting bases across China is under construction.

While popularizing the concept and practice of genetic counseling by organizing training courses and promoting model services, the CBGC has also completed the necessary paperwork to better guide the development of the profession. In its annual meeting in 2017, the CBGC published the first version of “China’s Expert Consensus on Guidance for Genetic Counseling.” The translation of the College of Medical Genetics and Genomics and the Association for Molecular Pathology (ACMG/AMP) guidelines (Richards et al., 2015; Wang et al., 2017) and the publication of the book "Genetic Counseling Today" by the community members demonstrated the leadership of the CBGC and collective efforts of the society. These works laid the essential foundation for the knowledge required for genetic counseling in China.

To better understand the genomes of Chinese population, particularly of those with significant birth defects or genetic diseases, the CBGC initiated a series of "Targeted Genomic Project" (http://www.
cbgc.org.cn). These projects aim to collect and sequence more than 10 k individuals within 3–5 years and create corresponding databases. The outcomes of these projects will play important roles in promoting proper genetic testing and genetic counseling.

2.4 | Challenges and directions for the future development of the genetic counseling profession in China

The effort for the rapid rise of the genetic counseling profession in China reflects the long unmet needs and the success of the CBGC’s promotion activities. Yet, there are a number of challenges that need to be addressed for eventual establishment of this profession in China.

First, the current CBGC training courses are designed to meet the most urgent needs by teaching general concepts and specific terms of medical genetics to participants of diverse background. The trainees include registered physicians, students enrolled in medical or life science schools, laboratory technicians, genetic teachers and researchers, as well as nurses who have worked as genetic counselors for some time. Balancing such a diverse group of academic backgrounds and satisfying all students in the same class can be difficult. More refined course structures need to be developed, including a structure that caters to the unique needs of physicians, laboratory technicians and genetic counselors.

Second, a comprehensive teaching materials and knowledgebase for genetic counseling is still lacking. Currently, an effort is under the way collecting real-world genetic counseling cases for Chinese patients. Formal teaching materials are being introduced, including those used for master degree education in the United States. Utilization of these resources should greatly improve course efficiency and consistency.

Third, students who graduated from the CBGC training courses are only receiving continuing education credits for their respective existing professions such as physicians and lab technicians. To make genetic counseling an allied health profession, the United States has developed the organizations managing the accreditation of training programs (Accreditation Council for Genetic Counseling, http://www.gceducation.org/), offering professional certification and continuous education (American Board of Genetic Counseling, ABGC, https://www.abgc.net/), and developing professional policies and practice guidelines (National Society of Genetic Counselors, https://www.nsgc.org/). We anticipate that a similar system will be put in place in future in China and most of genetic counselors will be trained in schools through a formal Master degree program, followed by a standardized clinical training system. The experiences accumulated in current CBGC training courses are paving the way to reach that stage.

Fourth, the development of genetic counseling profession in China needs to weave itself into the context of the development of this profession worldwide (Baty, 2018). Genetic counselors in developed countries have begun to address a greater number of clinical cases.
utilizing genomic data rather than targeted gene information, and deal
with cases in genetic conditions more complex than monogenic disor-
ders (Bowdin et al., 2016; Shelton & Whitcomb, 2015). Modern technol-
gies have also been employed to facilitate the process of counseling
(Gordon, Babu, & Laney, 2018). In addition, genetic counselors have
begun to work in more diverse settings, such as being a team member
of other medical specialty, in genetic testing laboratories, in research
departments, in commercial companies and with government agencies
(Middleton et al., 2017; Zetzsche, Kotzer, & Wain, 2014). In the United
States, genetic counselors may provide general service, or specialize in
one or more areas, including prenatal and preconception, pediatric, can-
cer, cardiovascular, neurology etc. Some genetic counselors also focus
on research. Due to these changes, the definition, the regulation, the
education system and the code of professional ethics of genetic coun-
selors continues to evolve, which must be considered when constructing
the professional system of genetic counseling in China (Hooker, Ormond, Sweet, & Biesecker, 2014; National Society of Genetic, 2018;
National Society of Genetic Counselors’ Definition Task et al., 2006).

Finally, public education in genomic science remains inadequate
in China. The general public is not at all primed to understand the
importance of genomic science to the health of the population and
individuals. Therefore, genetic counselors in China bear vital responsi-
bilities for public education and play critical roles in building communi-
cation between experts and laymen, conveying genetic knowledge and
cultivating a healthy market for genetic counseling services.

2.5 | Future outlook

On August 14, 2018, Shanghai Municipal Health Commission released a
governmental document titled “Management Measures of Genetic Counseling Technology Service in Shanghai (version 2018)” which offers
a clear definition of genetic counseling, details of educational and training
requirements for genetic counselors, and a precise description of a coun-
selor’s career duty and responsibilities. This document will play a vital role
for eventual establishment of the genetic counseling profession in China.

The development of genetic counseling profession today is a collective
effort by domestic and oversea experts, a synergistic result of the
increasing roles of medical genetics in modern medicine and the govern-
mental support for technology development. We expect that physicians
will integrate the genomic information into the process of disease diag-
nosis and treatment. Regulations, guidelines and standardized workflows
for genetic counseling will be issued by the government to facilitate this
process. Genomic databases of certain diseases will be well managed
and explored to clinical genetic screenings and diagnoses. Under these
circumstances, genetic counselors, will be well received in various
workplaces, including hospitals, governmental agencies, genetic testing
laboratories, research institutions and commercial companies (Biesecker,
2018). As medical genetics and precision medicine become a pivotal part
of modern medicine, the rise of genetic counseling profession is facilitating
the construction of a healthier China.

CONFLICT OF INTEREST

The authors declare that they have no conflict of interest. The submitted
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