Communication

On the Globalization and Standardization of Medical Genetics and Genomics as Clinical and Laboratory Specialties

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The evolving of medical genetics and genomics as a new specialty has been a major progress in the health care system during the past three decades. Since 2003, the completion of Human Genome Project has greatly speeded up the integration of genetics and genomics into mainstream clinical practice and thus demands more and more professionally trained medical genetics specialists. However, medical genetic training in different countries varies from well-organized professional system to lack of specialty recognition and shortage of specialists. These differences affect not only the quality of genetic services but also many related ethical, legal and social issues. To understand the current statues of medical genetics training in a global setting, we outline training systems from representative countries of North America, European Union, Latin American, Asia and Africa. The results demonstrate a trend toward formal recognition of medical genetics as a clinical specialty, diagnostic genetics as a laboratory specialty and genetic counseling as a Master level allied health profession. Systematic effort in training medical genetics specialists in developed countries has built up a workforce of one to seven clinical geneticists per million population. While in developing countries, lack of proper recognition and shortage of training programs are still the major obstacles for medical genetics. Considering the similar mutation rates and genetic disease burden on all human populations, globalization and standardization of medical genetics and genomics as clinical and laboratory specialties will contribute to better prevention and treatment of genetic diseases.

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INTRODUCTION

Medical genetics and genomics is the field for accurate diagnosis and effective prevention and treatment of human genetics diseases. It has been an integral part of current clinical practice in prenatal and postnatal diagnosis of inherited disorders and in differential diagnosis of somatic mutations for stratified treatment for many types of cancers. In the near future, the understanding of an individual's genome and its predisposing risks will be the foundation for personalized medicine. In 1971, medical genetics was first recognized as a certified specialization by Norwegian Medical Association.[1] From 1980s, many countries such as Portugal, Finland, United Kingdom, Netherlands, Germany, Austria, Czechoslovakia, Israel, Sweden, United States, Canada, Brazil and Argentina started to initiate training programs and then formally recognize this new medical specialty.[1-6] In 2011, clinical geneticists was officially recognized as an European Union (EU)-wide medical specialty and the accreditation of clinical laboratory geneticists is pending approval. This represents the first effort toward standardized medical genetics training across the EU countries. However, lack of specialty recognition, scarce of training programs and shortage of professionally trained clinical and laboratory geneticists are still the reality facing many developing countries including the two most populated nations China and India.[7-9] This report aims to outline the professional medical genetics training systems from representative countries around the world and present a reference for standardized medical genetics training in a global setting.

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MEDICAL GENETICS TRAINING IN NORTH AMERICA

In the United States, medical genetics specialty was initiated by the American Board of Medical Genetics and Genomics (ABMGG, http://www.abmgg.org) since 1982.[2,3] The ABMGG has accredited training programs and issued

specialty certifications for physician medical geneticists and laboratory specialists in cytogenetics, biochemical genetics and molecular genetics. Recently, two subspecialty programs medical biochemical genetics by ABMGG and molecular genetics pathology jointly by American Board of Pathology and ABMGG have also been developed. In 1991, the American Board of Medical Specialties (ABMS, http://www.abms.org) has approved the certification of MD for clinical genetics and MD / PhD for clinical cytogenetics, biochemical genetics, and molecular genetics. At the same time, the American Board of Genetic Counseling (ABGC, http://www.abgc.net) was established for accreditation of graduate level training programs and certification of master level genetic counselors. As of July 2014, there are 46 clinical genetics programs listed by the Accreditation Council for Graduate Medical Education (ACGME) as well as 44 clinical cytogenetics programs, 28 clinical biochemical genetics programs and 41 clinical molecular genetics programs listed in the ABMGG. There are 31 accredited programs for master-level genetic counselor listed in ABGC.

All physician residents and laboratory fellows require a two-year full time training for one specialty. Training for one additional specialty will need another one-year full time training. Residence programs combined medical genetics with internal medicine, maternal fetal medicine and pediatrics have been developed. Every training program will have its training manual with specified policies, procedures and guidelines. Generally speaking, laboratory fellows and physician residences are committed to case-oriented study and knowledge-based practice by a log book of 150 cases with a spectrum of representative genetic abnormalities. In-training exam for trainee evaluation has been implemented. Eligible clinical genetics residences and laboratory fellows passed the board exam will become certified specialists and diplomats of ABMGG.[4] As of July 2014, the ABMGG has listed a workforce consisting of 1,509 M.D. clinical geneticists, 315 clinical biochemical geneticists, 734 clinical cytogeneticists and 596 clinical molecular geneticists; additionally, there are 3,475 genetic counselors certified by ABGC. Based on a population size of 314 million and the pool of 1,500 clinical geneticists in the United States, there are about five clinical geneticists per million population. The ratio of clinical geneticists, laboratory specialists and genetic counselors is estimated to be 1:1:2. Steady growth in medical genetics workforce in USA has been obvious with newly-certified diplomats through biannual ABMGG board exams.

In Canada, medical genetics specialty was initiated by the incorporation of the Canadian College of Medical Geneticist (CCMG, http://www.ccmg-ccgm.org/) in 1975-76 and was recognized as a specialty within the Royal College in 1990. Physicians interested in becoming a clinical geneticist will take a three-year fellowship training at CCMG-accredited centers or a five-year direct-entry training program after medical school. Fellows with a doctoral degree (either PhD, MD, or equivalent) for laboratory geneticists will participate in a two-year postdoctoral training programs in biochemical genetics, cytogenetics and molecular genetics. There are nine training sites accredited by CCMG. The CCMG also recognizes ABMGG specialty certification as evidence of successful completion of CCMG-accredited training. From 1995 to 2013, certified clinical geneticists per million population increased from 0.8 to 2.6. Training to be a genetic counsellor is through a M.Sc. degree, offered at three Canadian Universities with certification by the Canadian Association of Genetic Counsellors (CAGC, http://www.cagc-accg.ca).

MEDICAL GENETICS TRAINING IN EUROPEAN UNION

In 1993, a survey by European Society of Human Genetics (ESHG, https://www.eshg.org/) found that clinical genetics is officially recognized in only four European Community (EC) countries with a total of 171 specialists (Germany, the Netherlands, Portugal, and UK) and in six non-EC countries (Austria, Czechoslovakia, Finland, Israel, Norway, and Sweden) with 119 specialists. Norway was the first in recognizing medical genetics specialty since 1971 and other countries recognized clinical genetics from 1979 to 1992. The training requirement varies from two-year residency (Germany) to five-year programs (Finland and Norway). The United Kingdom set an overall national target for two clinical geneticists per million population based on the recommendations of Royal College of Physicians of London. The survey presented wide discrepancy in status and training of clinical geneticists and urged other countries giving official recognition of medical genetics and favoring common European diplomas in medical genetics.[1]

Recently, the ESHG and the newly formed European Board of Medical Genetics (EBMG, 2012) have been working on EU-wide specialty in clinical genetics and laboratory genetics. In 2011, clinical genetics specialty was officially recognized as an EU-wide medical specialty. The national training curricula may vary from country to country but the training period should minimum four years full time work and extend for part time work. The ESHG recommended three clinical geneticists per million population. Current statistics showed a range of one to seven clinical geneticists per million populations in different EU countries; countries with pre-existed training programs like Germany, Sweden Czech Republic, Norway and Finland have 3.5 to 6 clinical geneticists per million population. An ESHG ad hoc laboratory genetics committee has been working on an EU-wide accreditation of clinical laboratory geneticists that joints cytogenetics, oncogenetics, molecular genetics and metabolic/biochemical genetics into one skilled profession. The entry level is M.Sc. or M.D. and the training period is also minimum four years full time work. The EBMG also proposes that all genetic counselors should have a Master degree in genetic counseling and genetic nurses a Master degree in genetic nursing.

MEDICAL GENETICS TRAINING IN LATIN AMERICA

In Latin America, it is estimated there are approximately 500 trained clinical geneticists, providing an array of medical genetic services. Genetic counseling is considered a medical function and is performed by clinical geneticists and other

trained physicians. The specialty of medical genetics has been recognized in Mexico, Cuba, Brazil and Argentina. However, residence training programs in clinical genetics are scarce and almost no country in the regions trains non-physician-allied health professionals to perform genetic counseling.[5,6]

In Brazil, the first medical residency program in Medical Genetics in University of Sao Paulo was recognized by the Ministry of Education in 1977. Medical Genetics was recognized as a medical specialty by the Federal Council of Medicine in 1983. Currently, there are 11 training programs and vacancies for 23 new physician trainees yearly. Board certification exams in Medical Genetics are held annually since 1993 by the Brazilian Society of Medical Genetics (BSMG) and involve a theoretical test, analysis of curriculum and interviews. Clinical geneticists can obtain specific certification in Medical Cytogenetics, Medical Molecular Genetics and Medical Biochemistry. Data collected in 2010 by the Brazilian Medical Association and the National Commission of Medical Residency shows that around 200 physicians have been awarded board certifications in Medical Genetics since 1981, which translates to about one clinical geneticist per million population.[10]

Since 1980s, Argentina counts on a single accredited four-year residence program in medical genetics for physicians. The program is located in the City of Buenos Aires and run by the National Medical Genetics Center of the Ministry of Health with two to three graduates per year. The specialty of medical genetics is recognized by the Ministry of Health since 1991. The Argentine Society of Genetics, in conjunction with the Ministry of Health, has certified about 120 geneticists who staffed 41 clinical genetics units throughout the country. There are about three clinical geneticists per million population. Genetics counseling is the responsibility of clinical geneticists in Argentina.[11]

MEDICAL GENETICS TRAINING IN ASIA

In Philippines, medical genetics is a recognized specialty since year 2000. The Department of Pediatrics at Philippine General Hospital offers a two-year fellowship program in Clinical Genetics. Components of the training program include broad clinical exposure to areas of dysmorphology, metabolic disorders, biochemical genetics, cytogenetics, molecular genetics and neonatal screening programs. A two-year Master of Science program in genetic counseling started in 2011, catering to nurses, doctors and other allied health professionals. In general, the field of clinical genetics is not a popular profession at this time.[12]

Medical genetics has not been recognized as a clinical specialty in Japan. However, the College of Laboratory Medicine of Japan in collaboration with the Japanese Society of Laboratory Medicine have developed a certification examination for technological professionals for the purpose of nurturing professionals possessing a high level of competency in the field of molecular analysis or molecular genetic testing, and thus providing the quality health care services. Certifications are separated into two levels: molecular analysis technologist and specialist. The former measures basic knowledge and skills. The latter measures the competencies defined in the statement, which includes compliance with the standards or guidelines for quality assurance of molecular genetic testing. The former began in 2007, and 398 professionals have been certified in 5 years. The latter began in 2012. Personnel qualification linked with the standards is expected to be an efficient and effective approach to providing the quality service.[13]

China has the largest population in the world and thus the biggest pool of genetic diseases. There are increasing demands and rapid growth of genetic testing services. However, medical genetics has not been recognized as a clinical specialty and there are no formal programs to train clinical genetics and laboratory professionals.[7] Since 1980, medical genetics centers and departments focusing on teaching genetic courses for medical students and performing research projects on inherited diseases have been structured in most of the medical schools. Institutional and hospital-based genetic services are increasing dramatically and genetic counseling training programs have been initiated.[14-17] In Taiwan, starting in 1992, clinical genetics training program was established as a two-year fellowship after one acquires a pediatric board certification. The first board examination was held in 1996. All trainees need to learn the clinical care of patients with genetic and metabolic conditions and also laboratory techniques of cytogenetics, molecular and biochemical genetics.[18]

In India, medical genetics exists as a super-specialty course (D.M.) only at the Department of Medical Genetics, Sanjay Gandhi Postgraduate Institute of Medical Science. The duration of training is three years and intake is about one to two fellows each year. A fellowship program by the Society for Indian Academy of Medical Genetics (SIAMG) offers 18-month training in clinical cytogenetics and molecular genetics with one trainee per year. Recently, a university in Hyderabad has started a structured genetic counseling course. Other institutions are likely to offer similar courses in the future.[8,9]

MEDICAL GENETICS TRAINING IN AFRICA

In South Africa, medical genetics was initially recognized as a subspecialty. In 1999, nine medical geneticists were registered through a grandfather clause and subsequently nine more medical specialists undertook the newly introduced two-year medical genetics training and were registered as medical geneticists. In 2007, medical genetics was recognized as a primary specialty in medicine. A four-year specialist training toward a postgraduate MMed degree and Fellowship of the College or Medical Genetics is offered at four universities. Currently, there are 11 medical geneticists and 7 in-training registrars, indicating one medical geneticist for 4,450,000 individuals. Genetic counselling is a recognized and registered health profession with formal two-year full-time postgraduate training at the Master's degree level.[19]

Table 1. List of Representative Countries with Specialty Training for Medical Genetics and Genomics.

	USA	Canada	European Union (EU)	Brazil	South Africa
Specialty Recognition	Initiated 1982, recognized 1991	Initiated 1975-76, recognized 1990	EU-wide recognition 2011	Initiated 1977, recognized 1983	Registered in 1999, recognized 2007
Organization	ABMGG, ACMG	CCMG	ESHG, EBMG	BSMG	
Specialty					
Clinical Geneticist	M.D., two-year training	MD, three-year fellowship or five-year program	MD, MS, four-year training	MD	MMed, four-year training
Laboratory Geneticists	M.D. and/or Ph.D., two-year training, specialized in Clinical Cytogenetics, Biochemical Genetics, and Molecular Genetics	MD and/or PhD, two-year training, specialized in Clinical Cytogenetics, Biochemical Genetics, and Molecular Genetics	EU-wide specialty pending approval, four-year training combining cytogenetics, oncogenetics, biochemical genetics, and molecular genetics as one profession	(Clinical geneticists specialized on clinical cytogenetics, biochemical genetics, and molecular genetics)	N/A
Genetic counselor	Master-level graduate study, certification by ABGC	Master-level graduate study, certification by CAGC	Master-level genetic counselor or nurse	(Genetic counseling as a medical function of clinical geneticists)	Master-level genetic counselor or nurse
CGPM*	5	2.6	1~7	1	0.22
CG:LG:GC Ratio*	1:01:02	N/A	N/A	N/A	N/A

*CGPM: clinical geneticist per million population; CG:LG:GC ratio: Clinical Geneticists:Laboratory Geneticists:Genetic Counselors ratio.
Note: See text for profession organizations ABMGG, ACMG, CCMG, ESHG, EBMG, BSMG, ABGC and CAGC

DISCUSSIONS

The medical genetics training systems from representative countries are summarized in **Table 1**. Despite differences in the training period, medical genetics and genomics as clinical and laboratory specialties and genetic counseling as a Master level allied health profession are the current setting. Medical genetics specialties in USA and Canada are almost identical and mutually recognized; similar medical genetics training systems have been structured in other countries and EU. The co-existence of M.D. clinical geneticists for disease management and mostly Ph.D. laboratory geneticists for genetic diagnosis has been the unique structure for this newly emergent medical field. This reflects the reality that evidence-based management of patients with genetic disorders is heavily relied on the technology-driven laboratory infrastructure for genetic disease diagnosis.[20] Well-trained specialists, sophisticated instruments and supportive resources are required for ever increasing number of genome-wide high throughput genetic tests. For example, the validation of high-throughput next-generation sequencing for clinical diagnosis needs efforts from regional or even international collaboration and the interpretation of clinical significance from sequencing findings requires comprehensive bioinformatic resources.[21,22]

In the United States, the medical genetics specialty is a typical top-down designed system by ABMGG and ACMG. It has proven very effective and successful in providing professional genetic diagnosis and clinical management, developing medical genetics policies, guidelines and standards for laboratory operations and disease treatment, organizing training programs and delivering genetic education to other non-genetics health professional and the public.[3.4] Recently, new opportunities in medical genetics include whole exome or genomic sequencing, pharmacogenetic testing, testing for risk of cancer and other adult-onset disorders, expanded newborn screening and carrier screening programs, and treatment of both rare and common genetic disorders. However, the number of physicians who complete ABMGG certification has remained flat for recent years due to mostly a non-high-pay specialty at this stage and partly the lack of visibility. The medical genetics community has discussed ways to increase the 'pipeline' of medical genetics trainees and present competencies for the physician medical geneticist in the 21st century.[23] Despite some challenges in its visibility and attractancy, the United States' medical genetic training system serves as a good model for many other countries to develop their own medical genetics specialty.

Lack of official recognition of medical genetics as clinical specialty, shortage of trained personnel and laboratory infrastructure for genetic tests are the major problems in many developing countries.[7-9] In China, the booming economy has led to significant improvement in laboratory infrastructure, an increasing awareness of inherited genetic diseases and demands for better care for patients with genetic disorders.[14-16] However, medical genetics has not been recognized as clinical and laboratory specialties in China, which severely affect the financial support from health administrative, the construction of professional training system and the organization of a knowledge and skilled workforce.[7] Except for a few board-certified geneticists recruited from abroad, physicians, laboratory personnel and nurses practicing genetic diagnosis and counseling are largely based on short-term training.[16,17] This shortcoming has severe consequences on the practice of clinical genetics. In the education system, limited training results in lack of awareness of common genetic disorders for medical practitioners and the general public.[24] In the management level, there is no specialized geneticists to develop consensus, policies, guidelines and standards for genetics services. In the service front, inadequately trained medical

genetics practitioners and genetic counselors may not have sufficient knowledge and skills to provide safe and effective genetic services. Based on the ESHG recommendation of three clinical geneticists per million population, China will need at least 4,000 clinical geneticists and an equal amount of laboratory specialists. It is strongly recommended that the Chinese Society of Medical Genetics (CSMG) works on a top-down designed and forward-looking action plan toward official recognition of medical genetics as clinical and laboratory specialties and initiation of training programs.

All human populations face similar mutation rates from our genome and share the same burden of genetic diseases in our society. The promise of medical genetics and genomics to maintain health, facilitate diagnosis, and cure or mitigate disease is dependent on the skillful translation of genomic science into meaningful action in the clinic. As part of effort to overcome the barriers to implementing genomic discoveries within medical practice, an inter-Society Coordinating Committee for Physician education in genomics (ISCC) was organized in the spring of 2013. A framework of physician competencies in genomic medicine has been outlined.[25] In addition to this inter-Society coordination within a nation, an international effort is needed to apply same principle and practice to the diagnosis, treatment and prevention of human genetic diseases. The global medical genetics community can work together toward standardized medical genetics specialty through developing mutually recognized or exchanging programs to speed up training process for developing countries. An international effort to joint regional medical genetics and genomics societies for professional training on medical genetics and genomics will definitely improve the genetic services and be beneficial to the mankind.

CONFLICT OF INTEREST

None.

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