Progress and Perspective of Professional Training in Medical Genetics and Genomics: A Report of the Association of Chinese Geneticists in America

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Medical genetics and genomics is a newly developed clinical specialty built upon accumulated knowledge of human genetics and discoveries of thousands of genetic disorders. Since 1991, medical genetics has been recognized as one of the 24 medical specialties and the American Board of Medical Genetics (ABMG) has organized training programs and provided certifications in clinical genetics, cytogenetics, molecular genetics and biochemical genetics. To publicize and promote this specialty, a task force on professional medical genetics training has been organized by members of the Association of Chinese Geneticists in America (ACGA). This report was written by the task force to outline the principle and practice of professional medical genetics training and review the progress and impacts of medical genetics and genomics in North America. For the past twenty years, the American College of Medical Genetics and Genomics (ACMG) has built a professionally trained work force and developed many policies, guidelines and standards governing clinical services and laboratory diagnostics. Educational programs to improve the knowledge and literacy on medical genetics for other health professionals and general public have been introduced. A comparison of the economic impact between basic genomic research and diagnostic genetic service showed similar impact multipliers in creating jobs and revenues. Genetic and genomic medicine is still in a stage of early development and rapid expansion but has been the driver for transforming ‘diagnostics/counseling’ based disease treatment toward ‘predictive/preventive’ oriented health maintenance. This report is aimed to introduce professional medical genetics training to qualified physicians and graduate students and also facilitate medical genetics specialty in China through professional consultation and exchange training programs.


Key Words: specialty in medical genetics and genomics, core competencies, clinical and economic impact, career perspective

INTRODUCTION

Medical genetics is a newly-evolved specialty involving the analysis of genetic information for the diagnosis, treatment and prevention of hereditary disorders and for the predisposing genetic risks of common diseases.¹ Genetic information affects all of us and our families in unique ways. Such information could make a diagnosis of a known genetic syndrome or predict events that will occur years in the future. An increasing amount of clinically significant genetic defects ranging from large numerical and structural chromosomal abnormalities, pathogenic copy number variants, to single base pair mutations have been characterized. Constitutional genetic defects have been the causes of thousands of Mendelian disorders and hundreds of chromosomal and genomic syndromes; somatic genetic defects have been used in the diagnosis, treatment stratification and prognosis predication of many types of tumors. Genetic risk factors associated with many common diseases like coronary artery disease, stroke, diabetes and rheumatism have been studied. However, disease-causing mechanisms underlying many genetic defects need to be elucidated so that rational treatment and prevention approaches can be developed. To meet the need of clinical genetic service and translational genetic research in the United States, medical genetics...
professionals have been initiated and organized by the American Board of Medical Genetics (ABMG, http://www.abmg.org) and the American College of Medical Genetics (ACMG, http://www.acmg.net) since the early 1980s. Training programs were developed for certifications of physician medical geneticists and laboratory specialists in cytogenetics, biochemical and molecular genetics. The physician medical genetics residency has been recognized by the American Board of Medical Specialties (ABMS, http://www.abms.org) since 1991. Master-level genetic counselors are certified by American Board of Genetic Counseling (ABGC, http://www.abgc.net).

Rapid growth in medical genetics and genomics has made it increasingly an integral part of mainstream health care in the United States. In China, however, there had not been any uniform planning of clinical and laboratory genetics developments on a national level and systematic genetic counseling still seemed to be at an early stage of development.

Clinical genetics has not received due emphasis and medical genetics is still not recognized as one of the medical specialties. In the 2011 International Congress of Human Genetics at Montreal, a group of members of the Association of Chinese Geneticists in America (ACGA) proposed the formation of a task force on professional medical genetics education. This first report from the task force is aimed to: 1) outline the principles of professional medical genetics training, 2) introduce training programs to qualified physicians and graduate students interested in pursuing a career in genetic and genomic medicine, 3) advocate the creation of a medical genetics specialty in China through professional consultation, and 4) to promote exchange training programs and collaborative translational genetic and genomic research projects.

PROFESSIONAL MEDICAL GENETICS TRAINING

In the United States and Canada, genetic defects are detected in reference clinical laboratories directed by certified geneticists specialized in clinical cytogenetics, molecular genetics and biochemical genetics. The detected defects and associated clinical phenotypes or disease risks will be delivered to the patients, their family members and other health care providers by certified clinical geneticists and genetic counselors. To ensure and promote knowledge-based, ethically-safe and cost-effective medical genetic services, principles and core competencies to support professional medical genetics training for MD/PhD, MD and PhD have been developed in the United States, Canada and Europe.

Principles of Medical Genetics and Genomics

The medical genetics and genomics professionals follow the six core competencies for all clinical specialties by the Accreditation Council for Graduate Medical Education (ACGME, http://www.acgme.org):

1. Professionalism - Demonstrate professional responsibilities, adherence to ethical principles and sensitivity to diverse patient populations.
2. Patient Care and Procedural Skills - Provide compassionate, appropriate and effective treatment for health problems and to promote health.
3. Medical Knowledge - Demonstrate knowledge about established and evolving biomedical, clinical and cognate sciences and their application in patient care.
4. Practice-based Learning and Improvement - Able to evaluate patient care practices; appraise and assimilate scientific evidence to improve practice.
5. Interpersonal and Communication Skills - Demonstrate skills that result in effective information exchange and teaming with patients, their families and professional associates.
6. Systems-based Practice - Demonstrate awareness of, and responsibility to, larger context and systems of health care. Be able to call on system resources to provide optimal care.


Core Competences for Specialists in Medical Genetics

An ACMG working group reported overarching competencies and discipline-specific competencies for the physician medical geneticist in the 21st century. An expert group working under the auspices of the EurpGentest project and European Society of Human Genetics Education Committee also proposed a set of core competences to establish common standards for education and practice in genetics health care.

Clinical Geneticist (MD, MD/PhD)

The overarching competencies including:

1. Evaluate medical, social and family histories and physical findings for genetic disorders.
2. Order and interpret genetic tests and apply results to management and counseling.
3. Integrate multiple sources of information to quantify genetic risks for the family.
4. Apply effective communication for patients with or at risk for genetic disorders.
5. Apply genetic knowledge to formulate, implement and monitor a management plan.
6. Assess and participate in a translational research or clinical trial.
7. Apply knowledge of the core public health functions to patient care.
8. Provide patient care with sensitivity to the related ethical issues.
9. Provide counseling to individuals regarding the application of whole genome or whole exome sequencing.

Discipline-specific competencies for the physician medical geneticist on biochemical/metabolic, cancer, connective tissue, cardiovascular, deafness, dermatologic, dysmorphology, endocrine, gastrointestinal, hematologic, immunological, nephrologic, neurogenetic, ophthalmologic, prenatal/reproductive, psychiatric, pulmonary, skeletal diseases can be found from the ACMG website.
Clinical Molecular Geneticist (MD, MD/PhD, PhD)
1. Supervise and direct the operations of a clinical molecular genetics diagnostic laboratory, including all technical and quality assessment procedures.
2. Perform molecular genetic tests for the purposes of diagnosis, ascertainment of carrier status and predictive testing.
3. Have a broad knowledge of molecular biology and genetics and understand the etiology, pathogenesis, clinical manifestations and management of human genetic disorders.
4. Provide results, interpretation and follow up recommendations to health-care professionals.
5. Participate in clinical research and in the introduction of new methods.

Clinical Cytogeneticist (MD, MD/PhD, PhD)
1. Supervise and direct the operations of a clinical cytogenetic laboratory, including all technical and quality assessment procedures.
2. Work proficiently in the cultivation of cells for prenatal, postnatal and cancer chromosomal examination and processing of DNA for genome-wide copy number detection.
3. Have a broad knowledge in human cytogenetics and genomics and understand the chromosome heteromorphisms, constitutional and somatic abnormalities, and natural history of cytogenetic and genomic disorders.
4. Provide results, interpretation, and follow up recommendations to health-care professionals.
5. Participate in clinical research and in the introduction of new methods.

Clinical Biochemical Geneticist (MD, MD/PhD, PhD)
1. Supervise and direct the operations of a clinical biochemical laboratory, including all technical and quality assessment procedures.
2. Carry out biochemical screening and testing related to diagnosis and management of genetic disease and carrier states, especially related to inborn errors of metabolism.
3. Have a broad knowledge of clinical biochemistry and genetics and understand the etiology, pathogenesis, clinical manifestations and management of inborn errors of metabolism and inherited biochemical disorders.
4. Provide results, interpretation, and follow up recommendations to health-care professionals.
5. Participate in clinical research and in the introduction of new methods.

Medical Genetics Training Programs in North America
The Residency Review Committee of the Accreditation Council for Graduate Medical Education (ACGME) accredits medical genetics residency programs, as well as two subspecialty programs: Medical Biochemical Genetics and Molecular Genetic Pathology (the latter is a joint subspecialty with the American Board of Pathology). The ABMG accredits clinical laboratory training programs in the following medical genetics specialties: Clinical Biochemical Genetics, Clinical Cytogenetics, and Clinical Molecular Genetics. As of June 2012, there are 49 clinical genetics programs, 29 clinical biochemical genetics programs, 44 clinical cytogenetics programs, and 39 clinical molecular genetics programs in the United States. Contact information and application credentials can be found from the ABMG website. Similar training programs in Canada can be found from the Canadian College of Medical Geneticist website (CCMG, http://www.ccmg-ccgm.org/). There are ten training sites accredited by CCMG. There are 31 ABGC accredited graduate programs for genetic counseling in the United States and three in Canada (see http://www.abgc.net/Training_Program_Accreditation/Accredited_Programs.asp).

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 residents 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-service exam for trainee evaluation has been implemented.8 Eligible clinical genetics residents and laboratory fellows passed the board exam will become certified specialists and diplomates of ABMG.

THE IMPACTS OF GENETIC AND GENOMIC MEDICINE
The Human Genome Project (HGP), an international public project led by the United States and a complementary private program, represented the largest single undertaking in the history of biological science. The working drafts of the human genome sequence were published in special issues of Nature (February 15, 2001) and Science (February 16, 2001). On April 14, 2003, the Human Genome Project was declared officially complete. The first decade post-HGP has been marked with rapid development of high-throughput genomic technologies and their immediate applications to clinical practice.78 Genetic and genomic testing has made possible a transition from reactive medicine to personalized, predictive, preventive and participatory (P4) medicine as highlighted by Hood and Galas.7 The benefits of this P4 medicine are the capacities to detect disease at an earlier stage when it is easier and less expensive to treat effectively; to stratify patients into groups that enable the selection of optimal therapy; to reduce adverse drug reactions by more effective early assessment of individual drug responses; to improve the selection of new biochemical targets for drug discovery; to reduce the time, cost, and failure rate of clinical trials for new therapies; and to shift the emphasis in medicine from reaction to prevention and from disease to wellness. Advances in genetics, genomics and molecular biology have transformed ‘diagnostics/counseling’ based disease treatment of genetic medicine toward ‘predictive/preventive’ oriented health maintenance of genomic medicine. The clinical impacts from the use of genomic technologies and genomic data in...
healthcare delivery and the need for policy and guideline development to ensure timely and effective transition to genomic medicine have been discussed.\textsuperscript{10-12}

![Figure 1](image.png)

**Figure 1.** The number of ABMG certified genetic specialists and estimated number of certified members in ACGA.

**Promoting Standardized, Effective and Safe Genetic Services**

*Building up a workforce for medical genetics and genomics*

From 1991 to 2011, the ACMG has built a workforce consisting of 1,419 MD clinical geneticists, 295 clinical biochemical geneticists, 687 clinical cytogeneticists and 536 clinical molecular geneticists; additionally, there are 3,026 genetic counselors certified by ABGC (Figure 1). Of these certified genetic specialists, ACGA members account for approximately 2% of clinical geneticists, 5% of biochemical geneticists, 7% of cytogeneticists, and 10% of molecular geneticists. Based on a population size of 312,000,000 and the pool of 1,400 clinical geneticists in the United States, there is one clinical geneticist in an average of 223,000 people. The ratio of clinical genetic physicians, laboratory specialties and genetic counselors is estimated to be 1:1:2. Steady growth in medical genetics workforce in USA has been obvious by increased ACMG annual meeting attendees from 500 to over 1,800 in the past ten years.\textsuperscript{2} There are 49 academic medical genetics training programs in the USA and each program has 6-12 certified specialists depending on the size of its operation. It could be estimated that at least 10-20% of medical genetics specialists are working in an academic setting. These 300-600 medical genetics specialists are the driving force in performing state-of-the-art clinical and diagnostic genetics services, training medical genetics professionals, and conducting translational research in human genetics and genomics.

**Developing acmg policies, guidelines and standards**

To promote effective and safe genetic service in the United States, the ACMG has organized professional practice policies and guidelines, laboratory quality assurance and therapeutics committees working on practice policies, guidelines and standards. Since 1995, The ACMG has published 23 policy statements, 27 practice guidelines and 15 standards.\textsuperscript{2} The ACMG has also an expert group worked on standardized newborn screening by a uniform panel, act form and algorithm.\textsuperscript{13} Recently, the field of medical genetics has focused on improving the effectiveness of professional training and integrating genomic technologies into genetic evaluation.

**Providing medical genetics education and dealing with other related issues**

The ACMG’s education committee and maintenance of certification subcommittee have organized continuing medical education (CME) activities and maintenance of certification (MOC) program for all ABMG diplomats. The ABMG has also organized a bi-annual Genetics Review Course for eligible trainees to prepare for board exam. The ACMG has an online learning center provide on-demand resource of cutting edge educational programs including recorded content of short courses and selected session highlights from ACMG annual clinical genetics meeting. Furthermore, the ACMG has an economic committee and social, ethical and legal committees working on related issues. For example, the ACMG conducts bi-annual salary survey and present the salary report on the ACMG website.

The infrastructure of professional medical genetic training and its clinical impact on genetic clinic and diagnosis are shown in Figure 2. The ABMG and ACMG have put tremendous efforts and made significant progress to fulfill its mission statement of improving health through medical genetics by 1) define and promote excellence in the practice of medical genetics and genomics in the integration of translational research into practice, 2) promote and provide medical genetics and genomics education, 3) increase access to medical genetics and genomics services and integrate them into patient care, 4) advocate for and represent providers of medical genetics and genomics services and their patients, and 5) maintain structure and integrity of ACMG and its value to members and the public.
Figure 2. The clinical impact of ABMG and ACMG on medical genetics and genomics. CME, continuing medical education; MOC, maintenance of certificate; GIM, Genetics in Medicine—the official journal of ACMG.

Table 1. Economic impacts of the post-HGP Genomic Research vs Genetics and Genomic Testing in the USA (in Millions $).

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<tr>
<td></td>
<td>Jobs-Years</td>
<td>Personal Income</td>
</tr>
<tr>
<td>Direct Impacts</td>
<td>76,146</td>
<td>4,048.60</td>
</tr>
<tr>
<td>Indirect Impacts</td>
<td>31,730</td>
<td>1,670.30</td>
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<tr>
<td>Induced Impacts</td>
<td>64,787</td>
<td>2966.5</td>
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<tr>
<td>Total Impacts</td>
<td>172,663</td>
<td>8,685.50</td>
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<td>Impact Multiplier</td>
<td>2.27</td>
<td>2.12</td>
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Table 2. Current Medical Genetics Practice in USA and Estimated Need in China.

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<td></td>
<td></td>
<td></td>
<td>Clinical Geneticists</td>
<td>Lab Specialists</td>
</tr>
<tr>
<td>USA</td>
<td>312,000,000</td>
<td>202,800</td>
<td>14,447,100</td>
<td>1,500</td>
<td>1,500</td>
</tr>
<tr>
<td>China**</td>
<td>1,344,000,000</td>
<td>806,400</td>
<td>5,739,358</td>
<td>[6500]</td>
<td>[6500]</td>
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* Calculated by annual birth rate of 13/1000 (USA) and 12/1000 (China) times 5% prevalence of births having congenital disabilities
** All numbers (within [ ]) in China is estimated by population size and the output is estimated by GDP (gross domestic products)
Improving Knowledge of Genetics and Genomics for Public and Health Professionals

As genetic and genomic applications expand in a wider range of clinical settings from individual health care to public health surveillance, it is very important to improve the genetic literacy of the general public and the genetic knowledge of non-geneticist health professionals. Efforts to improve public knowledge of genetics and genomics come from all levels and directions. The World Health Organization (WHO) has a Human Genetics Programme (http://www.who.int/genomics/en/) with online resources for health professionals on disease classification and coding systems, genetics organization and societies and for patients and public on patient’s right and supportive services. The WHO expert groups also presented comprehensive reports on policy making and ethical, legal and social implications of medical genetics and genomics.\textsuperscript{e3}

Federal government has also developed numerous plans and programs to promote genetic and genomic services. For example, the United States Secretary for Health and Human Services advisory committee on heritable disorders in newborns and children recommended medical genetics and genomics education for maternal and child health primary care physicians.\textsuperscript{14} The Office of Genomics and Disease Prevention in the Centers for Disease Control and Prevention (CDC) contracted with the Institute of Medicine (IOM) to convene a committee on the implications of genomics for the public’s health. Major scientific and policy issues related to genomics and public health, major supports for and challenges to the translation of genetic research into population health benefits, and approaches for the integration of genomic information into strategies for promoting health and preventing disease have been examined and discussed.\textsuperscript{15}

State Departments of Public Health have also developed various genetic and genomic resources and action plans. For example, state population-based surveys have been conducted to evaluate the public awareness and use of direct-to-consumer personal genomic tests; and the results indicated public health agencies should play roles in surveillance, education, and policy development on direct-to-consumer genomic tests.\textsuperscript{16} Numerous education programs to improve genetics literacy for grade K-12, undergraduates, medical professionals and the general public have been assessed and implemented.\textsuperscript{17-20} Core curriculum of genetics for medical school students has been updated by the Association of Professors of Human and Medical Genetics (APHMG, http://www.aphmg.org/) and the American Society of Human Genetics (ASHG, http://www.ashg.org/).\textsuperscript{24} Core competences for general health practitioners, dentists, nurses and counselors have been outlined.\textsuperscript{3} Many professional organizations, including the ACMG and ASHG, have developed reach-out educational programs and accessible genetic resources for different health professionals and the general public.

Economic Impact of Genomic Research and Genetic Testing

Current integration of high-throughput genomic technologies into genetic and genomic medicine is built upon the completion of the Human Genome Project (HGP). Its profound impact in human health and medicine is just emerging and will be sustainable for a long time. The economic impact of the HGP was evaluated by Battelle Memorial Institute (Battelle) in 2011.\textsuperscript{e5} The federal government invested $3.8 billion ($5.6 billion in 2010 $) in the HGP through its completion in 2003. This investment was foundational in generating cumulative economic output of $796 billion from 1988-2010. Every $1 of federal HGP investment has contributed to the generation of $141 in the economy.

In 2012, Battelle prepared a report on ‘The Economic and Functional Impacts of Genetic and Genomic Clinical Laboratory Testing in the United States’ for the American Clinical Laboratory Association.\textsuperscript{e6} The genetic and genomic testing industry is responsible for generating more than 116,000 jobs, nearly $6 billion in personal income for workers, and $16.5 billion in national economic output. The direct, indirect and induced impacts on employment, personal income, output, state/local and federal tax revenue from the post-HGP genomic research and genetic/genomic testing are shown in Table 1. It is interesting to note that the federal/private-funded genomic research and service-based genetic and genomic testing have similar impact multipliers through all categories. This result indicates that federal, state, institutional and private funding on medical genetic and genomic services will have strong economic output and sustainable growth potential. From an economic standpoint, genetic and genomic testing is at a relatively early stage of its development and represents an expanding industry for the United States. The post-HGP biomedical research programs such as HapMap, ENCODE and VARIOME will provide many web-accessible clinical and bioinformatic databases and resources.\textsuperscript{2} Novel genomic technologies will be developed by continuous research and development investment. Technology-driven and evidence-based genetic and genomic medicine is expected to grow substantially in the near future.\textsuperscript{8,21,22}

PROSPECTIVE FOR MEDICAL GENETICS AND GENOMICS

The ABMG-organized professional medical genetics training programs and the ACMG-guided clinical genetic practice in the USA have been an effective system for a new specialty (Figure 2). However, the current ABMG-certified work force of 1419 clinical geneticists, 234 biochemical geneticists, 687 clinical cytogeneticists and 536 molecular geneticists faces great challenges. The estimated number of people per physician for clinical genetics is 223,000. In contrast, according to the 2012 physician specialty data book by the Association of American Medical Colleges, the number of people per physician for other specialties such as cardiology, obstetrics and gynecology, pediatrics and internal medicine were 14,164, 7,654, 5,568 and 2,834, respectively. Based on the birth rate of 13 per 1000 population by Centers of Disease Control and Prevention and prevalence of 3-5% of births having intellectual and developmental disabilities and multiple congenital anomalies, one clinical geneticist could
face 87 to 145 newborn patients and their direct relatives each year. Additionally, clinical geneticists have to handle increasing number of prenatal patients and cancer patients. There are urgent needs in recruiting more trainees in all medical genetics specialties to meet the growing demand for genetic services using high-throughput genomic technologies and bioinformatic tools and to conduct translational genetic and genomic research. Qualified physicians and graduate students interested in pursuing a career in genetic and genomic medicine are encouraged to look for detailed information from the ABMG online resources and to contact medical genetics programs for training opportunities.

China has the largest population and thus the largest pool of patients with genetic disorders. Based on the population size and gross domestic product (GDP), it is estimated that China will need to build a workforce of 13,000 medical genetics specialists and 13,000 genetics counselors, and this workforce will create approximately 500,000 related jobs and $7 billion business output in genetics and genomic testing alone (Table 2). Significant progress has been achieved in China’s medical genetics education and research. The Chinese Society of Medical Genetics (CSMG), a branch of the Chinese Medical Association (CMA), was founded in 1986. National medical genetics key laboratories and regional medical genetics centers have been established. Since 2002, funded by a five-year, $2 million grant from the Fogarty International Center at the National Institutes of Health (NIH), Johns Hopkins has established and conducted an international training program in genetics in conjunction with Peking Union Medical College and Peking University. There have been many institution-based, hospital-affiliated and commercial genetic diagnostic services provided by medical genetics practitioners throughout China. Unfortunately, despite continuous efforts and advocacy for professional medical genetic training in China, clinical genetics and laboratory genetic diagnostics are still not recognized as a medical specialty in China. The lack of medical genetics professionals in China will affect the quality of genetic service to patients and their families, the capacity to conduct translational research, and the ingenuity to develop effective educational programs, regulations and guidelines. Consequently, misinterpretation and unethical use of genetic information, unregulated diagnostic testing, and over-commercialized premature therapeutic intervention could potentially cause severe damage to the healthcare system.

Many ACGA specialists have made and will continue to make significant contributions to the genetic education and research in China. This task force recognizes the importance of professional medical genetics training for the coming era of genomic medicine and urges the development of a medical genetic specialty in China. The ACGA medical genetic specialists could work on advocating for an action plan of a medical genetics specialty in China through professional consultation, developing on-the-job exchange training programs, and participating in collaborative and translational genetic and genomic research.

ONLINE RESOURCES
Medical genetics websites
Accreditation Council for Graduate Medical Education, http://www.acgme.org/
American Board of Medical Genetics, http://www.abmg.org/
American Board of Medical Specialties, http://www.abms.org
American Board of Genetic Counseling, http://www.abgc.net
American College of Medical Genetics and Genomics, http://www.acmg.net
Canadian College of Medical Geneticists, http://www.ccmg-ccgm.org/

Educational materials

CONFLICT OF INTEREST
None.

NOTE
All authors are organizing members of a task force on professional medical genetics training of the Association of Chinese Geneticists in America (ACGA)

REFERENCES
6. Robin NH, Sutton VR, Caldwell J, Jackson J, Irons M, Demmer L and the members of the in-service exam subcommittees. The development


