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## **Association of Professors of Human and Medical Genetics/ American Society of Human Genetics MEDICAL SCHOOL CORE CURRICULUM IN GENETICS**

### *PREAMBLE*

Medical genetics is one of the most rapidly advancing fields of medicine, and molecular genetics is now integral to all aspects of biomedical science. Every physician who practices in the 21st century must have an in-depth knowledge of the principles of human genetics and their application to a wide variety of clinical problems. The American Society of Human Genetics and the Association of Professors of Human and Medical Genetics have developed this Medical School Core Curriculum to provide guidance to deans and curriculum committees regarding medical genetics knowledge, skills, and behaviors that all current medical students will need during their careers as physicians. Each medical school must find the best way to incorporate genetics teaching into its own curriculum, but some generalizations are possible:

- Medical genetics provides a unique perspective on function of the human body in health and disease; it is both a clinical specialty and a basic science. Medical genetics teaching must span the entire undergraduate medical school curriculum and continue into the postgraduate years.
- Medical genetics must be explicitly included in the curriculum. Although some aspects of medical genetics overlap with and may be taught by other disciplines, specific learning objectives in medical genetics need to be established.
- A well-qualified medical genetics specialist (or small committee of medical geneticists) should be given the authority and responsibility for implementing the genetics curriculum at each medical school. This responsibility should extend throughout the undergraduate medical curriculum and include involvement in all courses that deal with genetic principles or disorders.
- Medical genetics can be taught effectively by a variety of methods and in various formats. Problem-based learning is particularly well-suited to medical genetics because it involves integration of skills and knowledge from many fields. Genetics can also be taught in various clinical contexts and at different points in clinical training, depending on the particular circumstances at each school. Specific clinical examples are important, but the focus of the curriculum must be on medical genetic principles illustrated by the examples.

Given the rapid advance of medical genetics, this Core Curriculum is a work in progress. (The previous version was published in the American Journal of Human Genetics (1995))

56:535-537.) The American Society of Human Genetics and the Association of Professors of Human and Medical Genetics welcome all comments on these objectives, which will be revised as necessary to reflect changes that occur in our understanding of genetics and its application to medicine.

### *GENERAL MEDICAL COMPETENCIES ESSENTIAL TO MEDICAL GENETICS*

During their training, medical students must acquire many general skills and behaviors that are important in all aspects of clinical practice, including medical genetics. These general competencies include the ability of students to:

- 1.1 explain the importance of disease prediction and prevention;
- 1.2 understand the developmental stages of human behavior, maturation, and intelligence;
- 1.3 apply appropriate techniques for conveying difficult medical information;
- 1.4 understand how to respond appropriately to patients' defense mechanisms;
- 1.5 recognize the importance of reiterating information to patients who are anxious or unfamiliar with the concepts being presented;
- 1.6 recognize the importance of patient confidentiality;
- 1.7 make appropriate referrals to genetics support groups, community groups, or other resources that can benefit the patient and family;
- 1.8 respect the autonomy of all patients, but also provide guidance with decision-making when requested;
- 1.9 respect patients' religious, cultural, social, and ethical beliefs, even if they differ from their own beliefs;
- 1.10 interpret their own attitudes toward ethical, social, cultural, religious and ethnic issues and develop an ability to individualize each patient or family member;
- 1.11 cope emotionally with patient responses;
- 1.12 recognize the limitations of their own skills and seek consultation when necessary;
- 1.13 effectively use resources such as medical textbooks, research articles, and computer-based systems to obtain information necessary for good patient care;

- 1.14 apply the principles of evidence-based medicine to clinical practice;
- 1.15 understand how clinical observations can provide insight into human biology and disease pathogenesis and, through research, lead to improvements in health; and
- 1.16 undertake a program of life-long learning.

### *SPECIFIC KNOWLEDGE REQUIREMENTS*

The practice of modern medicine includes recognition of the role of genetic factors in health and disease. Students must know:

#### **2.1 Structure and Function of Genes and the General Organization of the Human Genome**

- 2.1.1 what genes are, how they are organized and controlled, what they do, and how they segregate;
- 2.1.2 how gene expression is affected by differences in coding and non-coding regions, effects of trans-acting factors, and the structure of chromatin;
- 2.1.3 how protein function is influenced by mRNA and polypeptide processing and interactions;
- 2.1.4 how gene activity varies during development and in normal and pathological cell function;
- 2.1.5 what information can and cannot be predicted from the DNA sequence of a gene;
- 2.1.6 what information can be obtained from measuring RNA or protein levels that cannot be obtained from the DNA sequence alone;
- 2.1.7 how processes such as gene duplication and divergence, exon shuffling, and the activity of transposable elements help to explain genomic variability, redundancy, and plasticity;

#### **2.2 Genes and Disease**

- 2.2.1 the patterns of inheritance characteristic of autosomal dominant, autosomal recessive, X-linked dominant, and X-linked recessive traits;

- 2.2.2 factors that affect development of the phenotype in single-gene disorders, including modifier genes, and stochastic and pleiotropic effects, which result in variable expressivity and incomplete penetrance;
- 2.2.3 the clinical manifestations of common mendelian diseases;
- 2.2.4 the basic principles of inborn errors of metabolism and of pharmacogenetic variations and their general clinical manifestations;
- 2.2.5 the genetic basis of mitochondrial diseases and the expected inheritance patterns for mitochondrial traits;
- 2.2.6 the nature of mutations and premutations and how they contribute to human variability and disease;
- 2.2.7 the concepts and clinical importance of genetic imprinting and uniparental disomy;
- 2.2.8 how polymorphisms, human gene mapping, and gene linkage and association studies are used in medicine;
- 2.2.9 the multifactorial nature of most human traits, both normal and abnormal, and the principles of multifactorial inheritance;
- 2.2.10 how genes interact with other genes and with various environmental factors to produce disease, and how amelioration of non-genetic factors can prevent development of disease in a genetically-predisposed individual;

## **2.3 Chromosomes and Chromosomal Abnormalities**

- 2.3.1 how genes are organized into chromosomes, how chromosomes replicate in mitosis and meiosis, and how they are transmitted from parent to child;
- 2.3.2 the clinical features of common numerical, structural, and mosaic chromosomal abnormalities;

## **2.4 Population Genetics**

- 2.4.1 how the principles of population genetics account for varying frequencies of particular mutations in populations, the effects of consanguinity, the continuing occurrence of new mutations, and the resistance of gene frequencies to change by medical intervention;

- 2.4.2 how evolutionary principles can be used to understand human biology and disease;

## **2.5 Genetics in Medical Practice**

- 2.5.1 how knowledge of a patient's genotype can be used to develop a more effective approach to health maintenance, disease prevention, disease diagnosis, and treatment for that particular individual;
- 2.5.2 common molecular and cytogenetic diagnostic techniques and how they are applied to genetic disorders;
- 2.5.3 how constitutional and acquired genetic alterations can lead to the development of malignant neoplasms and how identification of these changes can be used in the diagnosis, management and prevention of malignancy;
- 2.5.4 the potential advantages, limitations, and disadvantages of presymptomatic testing for genetic disease;
- 2.5.5 the potential advantages, limitations, and disadvantages of predictive testing for genetic disease;
- 2.5.6 how appropriate applications of genetic medicine can improve public health, and how to determine whether such interventions are warranted in a particular population;
- 2.5.7 the alternative approaches and goals of screening programs for genetic diseases in newborn infants, pregnant women, and other adults, and the ethical issues involved in justifying each program;
- 2.5.8 the existence of and justification for screening programs to detect genetic disease, and the difference between screening and more definitive testing;
- 2.5.9 conventional approaches to treatment of genetic diseases and the general status of gene-based therapies;
- 2.5.10 what exposures are likely to be teratogenic in humans and how such exposures can be prevented;
- 2.5.11 how to recognize and classify congenital anomalies and multiple congenital anomaly syndromes;
- 2.5.12 the purpose of genetic counseling;
- 2.5.13 when and how to refer individuals with a genetic disease or congenital anomaly to medical genetics specialists, and why referral is beneficial to the patients;
- 2.5.14 how novel scientific discoveries are evaluated in a clinical context and applied appropriately to the care of patients;
- 2.5.15 how legal and ethical issues related to genetics affect general medical practice;

- 2.5.16 how organizational and economic aspects of the health care system affect delivery of clinical genetic services;
- 2.5.17 what lessons the history of use and misuse of human genetics teach about the proper application of contemporary medical genetic knowledge.

### *SPECIFIC SKILLS*

Students must learn to synthesize factual material related to genetic diseases and to use this information to formulate an appropriate plan for diagnostic evaluation and patient management. They need the ability to:

- 3.1 elicit a comprehensive family medical history, construct an appropriate medical pedigree, and recognize patterns of inheritance and other signs suggestive of genetic disease in the family history;
- 3.2 recognize features in a patient's medical history, physical examination or laboratory investigations that suggest the presence of genetic disease;
- 3.3 identify patients with strong inherited predispositions to common diseases and facilitate appropriate assessment of other at-risk family members;
- 3.4 recognize and classify common congenital anomalies and patterns of anomalies;
- 3.5 recognize and initiate the evaluation of patients with inborn errors of metabolism;
- 3.6 interpret the results of common cytogenetic, molecular genetic, and biochemical genetic diagnostic techniques efficiently;
- 3.7 estimate recurrence risks for mendelian and multifactorial disorders in affected families;
- 3.8 use the information that a patient has a genetic predisposition for a particular disease to help reduce the risk of developing that disease or deal with it more effectively if it does develop;
- 3.9 describe appropriate techniques and approaches to providing genetic counseling for commonly-encountered genetic diseases;
- 3.10 communicate genetic information in a clear and non-directive manner that is suitable for individuals of different educational, socio-economic, ethnic and cultural backgrounds;

- 3.11 recognize and accept varying cultural, social, and religious attitudes in relation to issues such as contraception, abortion, parenting, and gender roles;
- 3.12 utilize community support services and agencies, in particular, support groups for genetic diseases, appropriately;
- 3.13 provide patients with access to diagnostic and predictive tests that are appropriate for the condition in their family and advise patients of the benefits, limitations, and risks of such tests;
- 3.14 work with a medical genetics specialist to develop a comprehensive plan for the evaluation and management of patients with genetic disease;
- 3.15 make available to patients with genetic diseases appropriate treatments, including dietary, pharmacological, enzyme-replacement, transplantation, and gene therapies, as well as anticipatory guidance regarding health screening practices specific to the diagnosis;
- 3.16 appreciate the important role of biomedical research and acquire skills that enable critical analysis of scientific developments.

### *SPECIFIC BEHAVIORS*

Students must learn to be sympathetic, nonjudgmental, and non-coercive counselors who recognize their own limitations and seek consultation whenever necessary. Students should:

- 4.1 present all relevant options fairly, accurately, and non-coercively;
- 4.2 be aware of the dilemmas posed by confidentiality when relatives are found to be at risk for a serious disease;
- 4.3 appreciate the implications that information regarding a genetic abnormality can have for a person's self-image, family relationships, and social status and that patients' reactions may differ depending on factors such as gender, age, culture, and education;
- 4.4 when appropriate, encourage patient participation in medical research provided the patient and/or family is fully informed and understands the risks and benefits of participation in terms of their own disease, treatment, and social context.

