Clinical objectives in medical genetics for undergraduate medical students

PREAMBLE

The Association of Professors of Human or Medical Genetics developed these objectives to define the knowledge, skills, and attitudes in clinical genetics that all medical students should achieve during the clinical phase of their education. These objectives complement those of the American Society of Human Genetics (ASHG) Medical School Core Curriculum in Genetics, which covers both basic science and clinical aspects of medical student education (*Am J Hum Genet* 1995;56:535–537).

We assume that students who are learning clinical medicine already possess an understanding of the basic principles of human genetics. Clinical training provides students with an opportunity to apply their understanding of these principles to the diagnosis, management, and prevention of human disease. Learning medical genetics is a process that should continue throughout a physician's career. Medical students should gain cognitive mastery of the objectives listed below during clinical training. Subsequent postgraduate training should provide appropriate experience in the application of these objectives in primary care or a particular medical specialty.

Medical genetics is a recognized clinical specialty as well as an extensive field of knowledge that provides a unique perspective on the function of the human body in health and disease. It is, therefore, essential that a well-qualified medical geneticist (or small committee of medical geneticists) be given the authority and responsibility for implementing the clinical genetics curriculum at each medical school. We recognize that curricula, resources, and personnel available for teaching medical genetics differ greatly among medical schools. The objectives included here can be taught in various clinical contexts and at different points in clinical training, depending on the particular circumstances at each school.

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

- appreciate the importance of disease prediction and prevention;
- understand the developmental stages of human behavior, maturation, and intelligence;
- recognize the importance of patient confidentiality;
- apply appropriate techniques for conveying difficult medical information;
- recognize and understand how to respond appropriately to patients' defense mechanisms;
- tolerate and encourage reiteration of information because of patient anxiety or unfamiliarity with the concepts being presented;
- respect patients' religious, cultural, social, and ethical beliefs even if they differ from the students' own beliefs;
- respect the autonomy of all patients while giving appropriate consideration to the difficulties certain handicapping conditions may pose for decision-making;
- make appropriate referrals to community or other resources that can benefit the patient and family;
- interpret their own attitudes toward ethical, social, cultural, religious, and ethnic issues and develop an ability to individualize each patient or family member;
- cope emotionally with patient responses;
- recognize the limitations of their own skills and seek consultation when necessary;
- effectively use resources such as medical textbooks, research articles, and computer-based systems to obtain information necessary for good patient care;
- apply the principles of evidence-based medicine to clinical practice;
- understand how clinical observations can provide insight into human biology and disease pathogenesis and, through research, lead to improvements in health; and
- undertake a program of life-long learning.

Medical genetics is one of the most rapidly advancing areas of medical practice. The knowledge of medical genetics that is necessary for medical practice is likely to be very different in 10 or 20 years than it is now. The Association of Professors welcomes all comments on these objectives, which we shall revise as necessary to reflect changes that occur in our understanding of genetics and its application to clinical medicine.

CLINICAL OBJECTIVES IN MEDICAL GENETICS

Students should know:

1.1 how genetic factors predispose to mendelian and multifactorial diseases and the implications of such predisposition for disease diagnosis, treatment, and prevention;
1.2 the clinical manifestations of common mendelian diseases;
1.3 the clinical features of common chromosomal aneuploidies and the signs generally associated with other kinds of chromosomal imbalance;
1.4 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;
1.5 how knowledge of a patient's genotype can be used to develop a more effective approach to health maintenance, disease diagnosis, and treatment for that particular individual;
1.6 the procedures that are generally employed for prenatal genetic diagnosis and the indications for such testing;
1.7 the advantages, limitations, and disadvantages of presymptomatic testing for genetic disease;
1.8 the existence of and justification for screening programs to detect genetic disease, and the difference between screening and more definitive testing;
1.9 the differences in goals and approach among screening programs for genetic diseases in newborn infants, pregnant women, and other adults;
1.10 conventional approaches to treatment of genetic diseases and the general status of gene-based therapies;
1.11 what exposures are likely to be teratogenic in humans and how such exposures can be prevented;
1.12 indications and appropriate methods for referral of individu-

als with a genetic disease or congenital anomaly to medical genetics specialists;

1.13 how novel scientific discoveries are evaluated in a clinical context and applied appropriately to the care of patients; and

1.14 how legal and ethical issues related to genetics affect general medical practice.

Students should be able to:

2.1 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;

2.2 recognize features in a patient's medical history, physical examination, or laboratory investigations that suggest the presence of genetic disease;

2.3 identify patients with strong inherited predispositions to common diseases and facilitate appropriate assessment of other at-risk family members;

2.4 recognize and classify common congenital anomalies and patterns of anomalies;

2.5 recognize and initiate the evaluation of patients with inborn errors of metabolism;

2.6 use common cytogenetic, molecular genetic, and biochemical genetic diagnostic techniques efficiently;

2.7 estimate recurrence risks within an affected family for diseases transmitted as uncomplicated mendelian or multifactorial traits;

2.8 use the information that a patient has a genetic predisposition for a particular disease to help him or her reduce the risk of developing that disease or deal with it more effectively if it does develop;

2.9 describe appropriate techniques and approaches to providing genetic counseling for commonly-encountered genetic diseases;

2.10 communicate genetic information in a manner that is suitable for individuals of various different educational and cultural backgrounds; and

2.11 work with a medical genetics specialist to develop a comprehensive plan for the evaluation and management of patients with genetic disease.

Association of Professors of Human Genetics, Clinical Objectives Task Force: Jan M. Friedman, MD, PhD (chair), Miriam Blitzer, PhD, Louis J. Elsas II, MD, Uta Francke, MD, and Huntington F. Willard, PhD

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