



## Recommended Curriculum Guidelines for Family Medicine Residents

# Medical Genetics

*This document was endorsed by the American Academy of Family Physicians (AAFP) and the Society of Teachers of Family Medicine (STFM), and was developed in cooperation with the University of Nevada Family Medicine Residency Program.*

### Introduction

This Curriculum Guideline defines a recommended training strategy for family medicine residents. Attitudes, knowledge and skills that are critical to family medicine should be attained through longitudinal experience that promotes educational competencies defined by the Accreditation Council for Graduate Medical Education (ACGME) <http://www.acgme.org>. The curriculum must include structured experience in several specified areas. Most of the resident's knowledge will be gained by caring for ambulatory patients who visit the family medicine center. Structured didactic lectures, conferences, journal clubs and workshops must be included in the curriculum with an emphasis on outcomes-oriented, evidence-based studies that delineate common and chronic diseases affecting patients of all ages. Targeted techniques of health promotion and disease prevention are hallmarks of family medicine. Appropriate referral patterns and provision of cost-effective care should also be part of the curriculum.

Program requirements specific to family medicine residencies may be found on the ACGME Web site. Current AAFP Curriculum Guidelines may be found online at <http://www.aafp.org/cg>. These guidelines are periodically updated and endorsed by the AAFP and, in many instances, other specialty societies as indicated on each guideline.

Each residency program is responsible for its own curriculum. ***This guideline provides a useful strategy to help residency programs form their curricula for educating family physicians.***

### Preamble

The world has witnessed publication of the complete sequence of the human genome, the growing use of microarray technology to refine treatment in selected cancers and the increasing application of pharmacogenomics to develop new drugs. The volume of new information available to health care providers is transforming our understanding of

disease processes. The general public, empowered by access to information on the internet, grows better informed each day about genetics and genetically-based health care. Understanding the role genetics plays in health and disease provides the means to integrate the evolution of scientific discoveries from the study of genetics into diagnosis, prevention, treatment of many common diseases and an improvement in the health of society. The competencies related to genetics will encourage the integration of genetics knowledge, skills and attitudes into routine health care.

This Curriculum Guideline provides an outline of the attitudes, knowledge and skills that should be among the objectives of training programs in family medicine and which will lead to optimal incorporation of medical genetics into the care of patients by future family medicine physicians.

## **Competencies**

At the completion of residency training, a family medicine resident should:

- Be able to identify and provide all aspects of pretest and counseling to patients whose personal and/or family history indicates risk of genetic disorders. (Medical Knowledge, Communication)
- Be able to effectively interview patients in order to obtain relevant genetic environmental and behavioral risk factors. (Patient Care, Communication)
- Understand the ethical and social implications of genetic testing. (Medical Knowledge, Systems-based Practice)
- Recognize his or her own practice implications and seek consultation with other medical genetics health care providers. (Practice-based Learning and Improvement)

## **Attitudes**

The resident should develop attitudes that encompass:

- Recognition of the philosophical, theological, cultural and ethical perspectives influencing use of genetic information and services
- An appreciation for the sensitivity of genetic information and the need for privacy and confidentiality while delivering genetic education and counseling fairly, accurately and without coercion or personal bias. Sensitivity to the patients' and families' culture, knowledge and language level is also important.
- Recognition of the importance of the family physician, the medical geneticist and the genetics team as collaborators in the evaluation, diagnosis and management of patients referred for genetic consultation.
- Recognition of limitations of his or her own genetics expertise.
- Recognition of ethical, social, cultural, religious and ethnic issues and when personal values and biases in that regard may affect or interfere with care provided to patients.

## Knowledge

In the appropriate setting, the resident should demonstrate the ability to apply knowledge of:

1. Basic human genetics terminology, principles of human and medical genetics and basic patterns of biological inheritance and variation (both within families and within populations).
2. The importance of the three generation family history in assessing predisposition to disease, highlighting the difference between a pedigree and genogram.
3. The role of genetic factors in health maintenance and disease prevention.
4. The difference between clinical diagnosis of disease and identification of genetic predisposition to disease.
5. The role of behavioral, social and environmental factors to modify or influence genetics in the manifestation of disease.
6. The influence of ethnicity, culture, related health beliefs and economics in diagnosis and in the ability of the patient to use genetic information and services.
7. The potential physical and/or psychosocial benefits, limitations and risks of genetic information for individuals in the context of the family and community.
8. The range of genetic approaches to treatment of disease (including pharmacogenomics and gene therapy).
9. The indications and resources for genetic testing and referral to genetic specialists.
10. The history of misuse of human genetic information (eugenics) and the ethical, legal and social issues related to genetic testing and recording of genetic information.
  - a. Screening for genetic abnormalities
  - b. Prenatal-preconception testing
  - c. Presymptomatic genetic testing
  - d. Carrier testing for genetic disorders
  - e. Confidentiality
  - f. Risk assessment
  - g. Responsibility to inform
  - h. Discrimination issues (e.g., insurance coverage, employment)
  - i. Informed consent
  - j. Paternity determinations

## **Skills**

In the appropriate setting, the resident should demonstrate the ability to independently perform or appropriately refer:

1. Gathering genetic family history information (including an appropriate multi-generational family history)
2. Identifying patients who would benefit from genetic services
3. Explaining basic concepts of probability, disease susceptibility and the influence of genetic factors in maintenance of health and development of disease
4. Seeking appropriate assistance from and referring to appropriate genetics experts and peer support resources
5. Obtaining credible, current information about genetics for self, patients and colleagues
6. Effectively using new information technologies to obtain current information about genetics
7. Educating others about patient-focused policy issues
8. Participating in professional and public education about genetics
9. Providing culturally appropriate information about the potential risks, benefits and limitations of genetic testing
10. Educating patients about the range of emotional effects they and/or family members may experience as a result of receiving genetic information
11. Safeguarding the privacy and confidentiality of the genetic information of their clients
12. Informing patients of potential limitations to maintaining privacy and confidentiality of genetic information
13. Educating patients about availability of genetic testing and/or treatment for conditions seen frequently in practice
14. Providing patients with an appropriate informed consent process to facilitate decision making related to genetic testing

## **Implementation**

Implementation of this curriculum should include longitudinal experience throughout residency training. Physicians who have expertise in medical genetics should be available for conferences and electives for resident physicians. A multidisciplinary approach coordinated by the family physician is an appropriate way of structuring teaching experiences in this area. Individual teaching and small group discussion will

help promote appropriate attitudes. The resident should be able to incorporate genetic diagnoses in the differential of common and complex diseases when examining patients of all ages.

## Resources

Robinson A, Linden M. Clinical Handbook of Genetics. 2<sup>nd</sup> ed. Boston, Ma.: Blackwell Scientific Publications, 1993.

Bennett RL. The Practical Guide to the Genetic Family History. New York, N.Y.: John Wiley, 1999.

Seashore MR, Wappner RS. Genetics in Primary Care & Clinical Medicine. Stamford, Ct.: Appleton & Lange, 1996.

Baker DL, Schuette JL, Uhlmann WR. A Guide to Genetic Counseling. New York, N.Y.: J. Wiley & Sons, 1998.

Greenwood Genetic Center. Counseling Aids for Geneticists. 4<sup>th</sup> ed. Greenwood Genetic Center, 1999.

Greenwood Genetic Center. Growth References: Third Trimester to Adulthood. 2<sup>nd</sup> ed. Greenwood Genetic Center, 1998.

## Web Sites

GeneTests

<http://www.geneclinics.org>

<http://www.genetests.org>

National Newborn Screening & Genetics Resource Center

[http://genes-r-us.uthscsa.edu/resources/genetics/primary\\_care.htm](http://genes-r-us.uthscsa.edu/resources/genetics/primary_care.htm)

Online Mendelian Inheritance in Man

<http://www.ncbi.nlm.nih.gov/sites/entrez?db=OMIM>

University of Kansas Medical Center

<http://www.kumc.edu/gec/geneinfo.html>

The Foundation for Blood Research (FBR)

<http://www.fbr.org>

Genetic Alliance

<http://www.geneticalliance.org>

New York Online Access to Health (NOAH)

<http://www.noah-health.org/index.html>

National Institutes of Health- Office of Rare Diseases  
<http://rarediseases.info.nih.gov>

National Organization for Rare Disorders  
<http://www.rarediseases.org>

March of Dimes - Resource Center  
<http://www.modimes.org>

National Coalition for Health Professional Education in Genetics  
<http://www.nchpeg.org>

National Society of Genetics Counselors  
<http://www.nsgc.org>

The Centers for Disease Control and Prevention- The Human Genome Epidemiology  
Network (HuGE Net)  
<http://www.cdc.gov/genetics/hugenet/default.htm>

The Council for Responsible Genetics  
<http://www.gene-watch.org/>

Greenwood Genetic Center  
[www.ggc.org](http://www.ggc.org)

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