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FAMILY PHYSICIANS

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Recommended Curriculum Guidelines for Family Medicine Residents

Medical Genetics

This document was endorsed by the American Academy of Family Physicians (AAFP).

Introduction

This Curriculum Guideline defines a recommended training strategy for family medicine residents. Attitudes, behaviors, 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), www.acgme.org. The family medicine curriculum must include structured experience in several specified areas. Much of the resident's knowledge will be gained by caring for ambulatory patients who visit the family medicine center, although additional experience gained in various other settings (e.g., an inpatient setting, a patient's home, a long-term care facility, the emergency department, the community) is critical for well-rounded residency training. The residents should be able to develop a skillset and apply their skills appropriately to all patient care settings.

Structured didactic lectures, conferences, journal clubs, and workshops must be included in the curriculum to supplement experiential learning, with an emphasis on outcomes-oriented, evidence-based studies that delineate common diseases affecting patients of all ages. Patient-centered care, and targeted techniques of health promotion and disease prevention are hallmarks of family medicine and should be integrated in all settings. Appropriate referral patterns, transitions of care, and the 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 website. Current AAFP Curriculum Guidelines may be found online at 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.

Please note that the term “manage” occurs frequently in AAFP Curriculum Guidelines. “Manage” is used in a broad sense to indicate that the family physician takes responsibility for ensuring that optimal, complete care is provided to the patient. This does not necessarily mean that all aspects of care need to be directly delivered personally by the family physician. Management may include appropriate referral to other health care providers, including other specialists, for evaluation and treatment.

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 field of medical genetics is growing and advancing rapidly. From the publication of the human genome to individualized genetics data, the world has seen 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 professionals at a dramatically lowered cost has expanded exponentially and 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, and 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 will lead to optimal incorporation of medical genetics into the care of patients by future family physicians.

Competencies

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

- Perform an appropriate multigenerational family history and identify patients who have a personal medical condition and/or family history that indicates the risk of a genetically linked disorder, and provide appropriate counseling (Medical Knowledge, Interpersonal and Communication Skills)
- Effectively interview patients in order to obtain information about relevant genetic, environmental, and behavioral risk factors (Patient Care, Interpersonal and Communication Skills)

- Understand the important social, behavioral, and psychological implications health-related genetic information can have for individuals and families (Medical Knowledge, Systems-based Practice)
- Recognize his or her limitations and seek consultation with other medical genetics health care professionals as needed (Practice-based Learning and Improvement)
- Properly use family medical history tools and/or genetics pedigree charts, including a three-generation family history (e.g., www.hhs.gov/familyhistory/) (Patient Care, Medical Knowledge, Interpersonal and Communication Skills)

Attitudes and Behaviors

The resident should demonstrate attitudes and behaviors that encompass:

- Recognition of the philosophical, theological, cultural, and bioethical perspectives influencing use of genetic information and services
- Appreciation for the sensitivity of genetic information, and the need for privacy and confidentiality while delivering genetic education and counseling fairly and accurately, without coercion or personal bias, and with sensitivity to the patients' and families' culture, knowledge, and language level
- Recognition of the importance of the family physician, the medical geneticist, and the genetics team as collaborators in the evaluation, diagnosis, and treatment of patients tested and referred for genetic consultation
- Recognition of ethical, social, cultural, religious, and ethnic issues and situations in which personal values and biases pertaining to these issues may affect or interfere with care provided to patients

Knowledge

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

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 that modify or influence genetics in the manifestation of disease
6. The influence of ethnicity, culture, related health beliefs, and economics in determining the ability of the patient to use genetic information and services
7. The potential physical and/or psychosocial benefits and risks of genetic information for individuals in the context of the family and community
8. The implementation of relevant practice guidelines or consensus statements
9. The range of genetic approaches to treatment of disease (including pharmacogenomics and gene therapy)
10. The indications and resources for genetic testing and referral to genetic specialists
11. The history of misuse of human genetic information (i.e., eugenics)
12. The ethical, legal, and social issues related to genetic testing and recording of genetic information regarding:
 - a. Screening for genetic abnormalities
 - b. Prenatal/preconception testing
 - c. Presymptomatic genetic testing
 - d. Carrier testing for genetic disorders
 - e. Confidentiality (Genetic Information Nondiscrimination Act of 2008 [GINA])
 - 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 the following:

1. Gather genetic family history information (including an appropriate multi-generational family history)
2. Identify patients who would benefit from genetic services

3. Explain basic concepts of probability, disease susceptibility, and the influence of genetic factors on maintenance of health and development of disease
4. Appropriately seek assistance from and refer to genetics experts and peer support resources
5. Obtain current information about genetics for self, patients, and colleagues
6. Utilize new information technologies effectively to obtain current information about genetics
7. Participate in professional and public educational discussions about genetics from the perspective of a family physician
8. Provide culturally appropriate information about the potential risks, benefits, and limitations of genetic testing
9. Educate patients about the range of emotions they and/or family members may experience as a result of receiving genetic information
10. Safeguard the privacy and confidentiality of the genetic information of patients
11. Inform patients of potential limitations of maintaining privacy and confidentiality of genetic information
12. Educate patients about availability of genetic testing and/or treatment for conditions seen frequently in practice, such as the following:
 - a. Breast Cancer genes 1 and 2 (BRCA1/BRCA2) testing for hereditary breast and ovarian cancer
 - b. Fluorescence in situ hybridization (FISH) analysis in the newborn nursery
 - c. Preconception counseling for carrier disease states
 - d. Risk scoring and focused testing for disease states (e.g., cancers that might or might not respond to treatments such as chemotherapy)
 - e. Screening for cardiogenetic anomalies, such as hypertrophic cardiomyopathies, lipid genetics, and coronary artery disease (CAD) risk states, as well as long QT syndrome, if appropriate
13. Provide patients with an appropriate informed consent process to facilitate decision making related to genetic testing
14. Educate patients about direct-to-consumer (DTC) genome-wide association study (GWAS) testing as a risk-stratification strategy, and its benefits and risks
15. Utilize appropriate genetic and genomic information resources

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. Consideration of genetic diagnoses in the differential diagnosis of common and complex diseases should be made during obstetrics, pediatric, and adult medicine training rotations, as well as during patient care in the family medicine center.

Resources

Bennett RL. *The Practical Guide to the Genetic Family History*. 2nd ed. New York, NY: Wiley-Blackwell; 2010.

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Skirton H, Patch C. *Genetics for Healthcare Professionals: A Lifestage Approach*. London, UK: Taylor & Francis; 2002.

Vamur H. Ten years on—the human genome and medicine. *N Engl J Med*. 2010;362(21):2028-2029.

Website Resources

American Board of Medical Genetics and Genomics. <http://abmgg.org/>

American College of Medical Genetics and Genomics. www.acmg.net

Centers for Disease Control and Prevention. Human Genome Epidemiology Network (HuGENet). www.cdc.gov/genomics/hugenet/default.htm

Centers for Disease Control and Prevention. Evaluation of Genomic Applications in Practice and Prevention. <https://www.cdc.gov/egapprovals/>

Genetic Alliance. www.geneticalliance.org

Genetics in Primary Care Institute.
<https://geneticsinprimarycare.aap.org/Pages/default.aspx>

March of Dimes. <http://www.marchofdimes.org/>

National Center for Advancing Translational Sciences (NCATS). U.S. Department of Health and Human Services, National Institutes of Health, Genetic and Rare Diseases Information Center. <https://rarediseases.info.nih.gov/>

National Coalition for Health Professional Education in Genetics (NCHPEG).
<https://www.jax.org/education-and-learning/clinical-and-continuing-education>

National Human Genome Research Institute. U.S. Department of Health and Human Services, National Institutes of Health. www.genome.gov/

National Newborn Screening & Global Resource Center (NNSGRC). Genetics in Primary Care (GPC) Training Program Curriculum Materials. http://genes-r-us.uthscsa.edu/resources/genetics/primary_care.htm

National Organization for Rare Disorders (NORD). <https://rarediseases.org/>

National Society of Genetics Counselors. www.nsgc.org

University of Kansas Medical Center. Information for Genetic Professionals.
www.kumc.edu/gec/geneinfo.html

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Medicine Rural Family Medicine Residency Program