



AMERICAN ACADEMY OF  
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](http://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 skill set 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](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 field of medical genetics is growing and advancing rapidly. From the publication of the human genome to individualized genetics data and at-home genetics testing, the world has seen the growing use of 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 lower cost has expanded exponentially and is transforming our understanding of disease processes. The general public 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 better diagnose, prevent, and treat many common diseases. Genetics, along with the fundamentals of family history gathering allows family physicians to accurately assess risk, counsel patients in preventive behaviors, and place genetic risk into its proper environmental and behavioral context. This holistic approach will impact the care of individuals and families longitudinally, even as new genetic knowledge is acquired.

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 to obtain information about relevant genetic, environmental, and behavioral risk factors (Patient Care, Interpersonal and Communication Skills)
- Understand the important psychosocial, behavioral, and ethical implications health-related genetic information can have for individuals and families (Medical Knowledge, Systems-based Practice)
- Recognize residents' limitations and seek consultation with other medical genetics health care professionals as needed (Practice-based Learning and Improvement, Systems-based Practice)
- Counsel patients on implications of genetic testing and results, and direct patients and families to appropriate patient-centered resources and/or specialists when

necessary (Patient Care, Medical Knowledge, Interpersonal and Communication Skills)

- Describe an individual's risk for disease, combining family history, genetics, and environmental and behavioral factors for both rare and common disorders. (Patient Care, Interpersonal and Communication Skills)

## **Attitudes and Behaviors**

The resident should develop attitudes 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 unbiased genetic education with sensitivity to the patients' and families' culture, knowledge, and language level
- Recognition of the importance of the genetics team as collaborators in the evaluation, diagnosis, and decision to pursue or forgo treatment for patients undergoing genetic consultation
- Recognition of ethical, social, cultural, religious, and ethnic issues and situations in which the provider's 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:

1. Basic human genetics principles and patterns of inheritance and variation (both within families and within populations)
2. The importance of the three-generation family history in assessing predisposition to disease
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 for individuals, families, and community
5. The role of behavioral, social, and environmental factors that modify or influence genetics in the manifestation of disease
6. The influence of culture, related health beliefs, and socioeconomic status in determining patient access to genetic information and services
7. The implementation of relevant practice guidelines or consensus statements

8. The range of genetic approaches to treat 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 (i.e., eugenics)
11. The ethical, legal, and social issues related to testing and recording of genetic information regarding:
  - a. Screening for genetic abnormalities
  - b. Prenatal/preconception testing
  - c. Presymptomatic genetic testing
  - d. Carrier testing
  - 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 consultation
3. Explain basic concepts of probability, disease susceptibility, and the influence of genetic and social 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. Consider genetic influence on all potentially relevant disease processes encountered
7. Provide unbiased information about the potential risks, benefits, and limitations of genetic testing

8. Educate patients about the range of emotions they and/or family members may experience as a result of receiving genetic information
9. Safeguard the privacy and confidentiality of the genetic information of patients and warn of potential limitations
10. Educate patients about availability of genetic testing and/or treatment for conditions seen frequently in practice, such as the following:
  - a. BReast CAncer (BRCA)1/BRCA2 testing for hereditary breast and ovarian cancer
  - b. Newborn screening
  - 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
  - f. Colon cancer
  - g. Prenatal testing (i.e. amniocentesis, quad screen, etc.)
  - h. Pharmacogenomics (i.e. with psychiatric medications)
11. Provide patients with an appropriate informed consent process to facilitate decision making related to genetic testing
12. Educate patients about risks and benefits with direct-to-consumer (DTC) genetic testing
13. Emerging diagnostic genetic testing – whole genome analysis

## **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 all training rotations, including obstetrics, pediatrics, and adult medicine in both inpatient and outpatient settings.

## **Resources**

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

Burke S, Martyn M, Stone A, Bennett C, Thomas H, Farndon P. Developing a curriculum statement based on clinical practice: genetics in primary care. *Br J Gen Pract*. 2009;59(559):99-103.

Feero WG, Guttmacher AE, Collins FS. Genomic medicine—an updated primer. *N Engl J Med*. 2010;362(21):2001-2011.

Greenwood Genetic Center. *Genetic Counseling Aids, 2013*. 6<sup>th</sup> ed. Greenwood, SC: Greenwood Genetic Center; 2013

LoPresti L, Levy HP. Genetics update. *FP Essentials™*, Edition No. 396. Leawood, KS: American Academy of Family Physicians; 2012.

Manolio TA. Genomewide association studies and assessment of the risk of disease. *N Engl J Med*. 2010;363(2):166-176.

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

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

U.S. Department of Health and Human Services. National Institutes of Health. National Institute of General Medical Sciences. The New Genetics. [www.nigms.nih.gov/education/Booklets/the-new-genetics/Documents/Booklet-The-New-Genetics.pdf](http://www.nigms.nih.gov/education/Booklets/the-new-genetics/Documents/Booklet-The-New-Genetics.pdf)

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

## **Website Resource**

### **For Providers**

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

American College of Medical Genetics and Genomics. [www.acmg.net](http://www.acmg.net)

Centers for Disease Control and Prevention. Population Research, Human Genome Epidemiology Network (HuGENet). [www.cdc.gov/genomics/hugenet/default.htm](http://www.cdc.gov/genomics/hugenet/default.htm)

Evaluation of Genomic Applications in Practice and Prevention. [www.egappreviews.org/](http://www.egappreviews.org/)

Genetic Alliance. [www.geneticalliance.org](http://www.geneticalliance.org)

Genetics in Primary Care Institute. [www.aap.org/en-us/advocacy-and-policy/aap-health-initiatives/Pages/Genetics-in-Primary-Care-Institute.aspx](http://www.aap.org/en-us/advocacy-and-policy/aap-health-initiatives/Pages/Genetics-in-Primary-Care-Institute.aspx)

National Coalition for Health Professional Education in Genetics (NCHPEG). [www.nchpeg.org](http://www.nchpeg.org)

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

National Society of Genetics Counselors. [www.nsgc.org](http://www.nsgc.org)

University of Kansas Medical Center. Information for Genetic Professionals. [www.kumc.edu/gec/geneinfo.html](http://www.kumc.edu/gec/geneinfo.html)

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

U.S. Department of Health and Human Services, National Institutes of Health, National Human Genome Research Institute. [www.genome.gov/](http://www.genome.gov/)

## **For Patients**

Centers for Disease Control and Prevention. Knowing is Not Enough – Act on Your Family Health history. [www.cdc.gov/genomics/famhistory/knowning\\_not\\_enough.htm?CDC\\_AA\\_refVal=https%3A%2F2Fwww.cdc.gov%2Ffeatures%2Ffamilyhealthhistory%2Findex.html](http://www.cdc.gov/genomics/famhistory/knowning_not_enough.htm?CDC_AA_refVal=https%3A%2F2Fwww.cdc.gov%2Ffeatures%2Ffamilyhealthhistory%2Findex.html)

March of Dimes. [www.marchofdimes.org/](http://www.marchofdimes.org/)

National Organization for Rare Disorders (NORD). [www.rarediseases.org](http://www.rarediseases.org)

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Revised 1/2004

Revised 3/2008

Revised 11/2009

Revised 6/2012 by Atlantic Health System

Revised 6/2014 by Akron General Center for Family Medicine, OH

Revised 12/2016 by University of Nevada School of Medicine's Department of Family and Community Medicine Rural Family Medicine Residency Program

Revised 8/2019 by Penn Medicine Lancaster General Family Medicine Residency Program