

Recommended Curriculum Guidelines for Family Medicine Residents

Medical Genetics

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 to improve the health of society. The competencies related to genetics will encourage the integration of genetics knowledge, skills, and attitudes into routine health care to provide effective care to individuals and families.

Attitudes

The resident should develop attitudes that encompass:

- A. Recognition of philosophical, theological, cultural, and ethical perspectives influencing use of genetic information and services
- B. 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 while being sensitive to the patients' and families' culture, knowledge and language level
- C. 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.
- D. Recognition of limitations of their own genetics expertise
- E. 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

The resident should demonstrate knowledge of:

- A. Basic principles of human and medical genetics
 1. Genes and chromosomes
 2. Genogram and pedigree
 3. Basic Mendelian inheritance patterns (hair/eye color, blood type)
 4. Non-Mendelian inheritance patterns
- B. Ethical and legal considerations/controversies
 1. Screening for genetic abnormalities
 2. Prenatal-preconception testing
3. Presymptomatic genetic testing (breast cancer genes, Huntington's disease)
4. Carrier testing for genetic disorders
5. Confidentiality
6. Risk assessment
7. Responsibility to inform
8. Discrimination issues (insurance coverage, employment)
9. Informed consent
10. Paternity determinations
- C. Terminology used in medical genetics (mosaicism, incomplete penetrance, variable expressivity, pleomorphic, malformation, deformation, disruption, dysmorphic, minor/major anomaly, homozygote, heterozygote, allele, polymorphism)
- D. Laboratory studies and research
 1. Karyotype
 2. Fluorescent in situ hybridization
 3. Polymerase chain reaction, sequencing, mutation detection
 4. Gene mapping
- E. Limitations of genetic testing (polymorphism versus mutation)
- F. The genetic implications of common disorders and conditions
 1. Chromosomal abnormalities
 2. Familial variants
 3. Oncology
 4. Geriatric disorders
 5. Metabolic disorders
 6. Skeletal/connective tissue abnormalities
 7. Cardiopulmonary
 8. Hematologic disorders
 9. Gastrointestinal abnormalities
 10. Neuromuscular disorders
 11. Neural tube defects
 12. Craniofacial abnormalities
 13. Psychiatric disorders
 14. Prenatal abnormalities (including carrier and maternal influence factors)
- G. Approach to the dysmorphic child/adult with multiple congenital abnormalities
- H. Common questions and misconceptions in genetic advances

Skills

The resident should demonstrate skills in:

- A. Gathering genetic family-history information, including an appropriate multi-generational family history
- B. Identifying patients who would benefit from genetic services
- C. Explaining basic concepts of probability and disease susceptibility, and the influence of genetic factors in maintenance of health and development of disease
- D. Seeking appropriate assistance from and refer to appropriate genetics experts and peer support resources
- E. Obtaining credible, current information about genetics, for self, patients, and colleagues
- F. Using effectively new information technologies to obtain current information about genetics
- G. Educating others about patient-focused policy issues
- H. Participating in professional and public education about genetics
- I. Providing appropriate information about the potential risks, benefits, and limitations of genetic testing
- J. Educating patients about the range of emotional effects they and/or family members may experience as a result of receiving genetic information
- K. Safeguarding privacy and confidentiality of genetic information of clients
- L. Informing patients of potential limitations to maintaining privacy and confidentiality of genetic information

The following are considered advanced skills and are not required of all residents:

- M. Educating patients about availability of genetic testing and/or treatment for conditions seen frequently in practice
- N. Providing patients with an appropriate informed consent process to facilitate decision making related to genetic testing

- O. Providing and encouraging use of, culturally appropriate, user-friendly materials/media to convey information about genetic concepts
- P. Explaining potential physical and psychosocial benefits and limitations of gene-based therapeutics for clients
- Q. Discussing costs of genetic services, benefits and potential risks of using health insurance for payment of genetic services, potential risks of discrimination ■

Resources

Web Sites

GeneTests-GeneClinics
<http://www.geneclinics.org> OR
<http://www.genetests.org>

Genetics in Primary Care: A Faculty Development Initiative (case studies)
http://genes-r-us.uthscsa.edu/resources/genetics/primary_care.htm
 Online Mendelian Inheritance in Man
<http://www3.ncbi.nlm.nih.gov/Omim>

Kansas University Medical Center
<http://www.kumc.edu/gec/geneinfo.html>

The Foundation for Blood Research (FBR)
<http://www.fbr.org>

The Genetics Resource Center
<http://www.pitt.edu/~edugene/resource>

Genetic Alliance
<http://www.geneticalliance.org>

New York Online Access to Health (NOAH)
<http://www.noah-health.org/index.html>
 Office of Rare Diseases
<http://rarediseases.info.nih.gov/ord/index.html>

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

Rare Genetic Diseases In Children
<http://mccr2.med.nyu.edu/murphp01/homenew.htm>

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

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

GROW (Genetics Resources on the Web) Search Tool
<http://search.info.nih.gov/grow>

HuGEM II Project
<http://www.dml.georgetown.edu/hugem>

American Society of Human Genetics
<http://www.faseb.org/genetics/ashg/ashgmenu.htm>

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

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/org.html>

Clinical Handbook of Genetics; Robinson A. And Linden, M: ISBN 0-86542-194-3

Practical Guide to the Genetic Family History; Bennett, R.: ISBN 0-471-25154-2

Genetics in Primary Care; Seashore, M. and Wappner, R.: ISBN 0-8385-3128-8

A Guide to Genetic Counseling, Baker, D., Schuette, J., and Uhlmann, W.: ISBN 0-471-18541-8

Counseling Aids for Geneticists, Greenwood Genetic Center (4th edition): 1-(888)-GGC-GENE www.ggc.org

Growth References: Third Trimester to Adulthood, Greenwood Genetic Center: ISSN 0733-124X

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