AMERICAN ACADEMY OF FAMILY PHYSICIANS

Recommended Curriculum Guidelines for Family Medicine Residents

Medical Genetics

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

Introduction

Each family medicine residency program is responsible for its own curriculum. The AAFP Commission on Education's Subcommittee on Graduate Curriculum has created this guide as an outline for curriculum development, and it should be tailored to the needs of the program. Through a series of structured and/or longitudinal experiences, the curricula below will support the overall achievement of the core educational competencies defined by the Accreditation Council for Graduate Medical Education and provide guideposts to program requirements specific to family medicine. For updates and details, please refer to the ACGME website at www.acgme.org. 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.

Preamble

Understanding the role genetics play in health and disease provides the means to better diagnose, prevent and treat many common diseases. A holistic approach will impact the care of individuals and families longitudinally, even as new genetic knowledge is acquired. Almost every child born in the United States undergoes state-mandated newborn screening. Genetic testing is routinely offered to women who have infertility. Additionally, 16% of patients seeking assisted reproductive technology in the United States reported preimplantation genetic testing as the reason for starting cycles. In the prenatal space, routine genetic testing is offered to all pregnant mothers.

This curriculum guideline outlines the objectives for training programs in family medicine and will lead to the optimal incorporation of medical genetics into the care of patients by future family physicians.

Patient Care

At the completion of residency, residents should be able to:

1. Identify and accurately describe phenotypic features or anomalies using standardized nomenclature
2. Recognize complex syndromes or disorders
3. Identify and access appropriate clinical guidelines to develop and implement plans for genetic screening, diagnosis and treatment, including but not limited to:
    a. Hereditary breast and ovarian cancer
    b. Hereditary nonpolyposis colorectal cancer
    c. Prostate cancer
    d. Sickle cell disease
    e. Thalassemia
    f. Congenital heart defects
    g. Phenylketonuria
    h. Cystic fibrosis
    i. Tay-Sachs disease
4. Perform an appropriate multigenerational family history
5. 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
6. Understand the important psychosocial, behavioral and ethical implications that health-related genetic information can have for individuals and families
7. Counsel patients on the implications of genetic testing and results, and direct patients and families to appropriate patient-centered resources and/or specialists when necessary
8. Understand the role of behavioral, social and environmental factors that modify or influence genetics in the manifestation of disease
9. Describe an individual's risk for disease, combining family history, genetics, environmental and behavioral factors for both rare and common disorders
10. Locate and apply the best available evidence integrated with patient preferences
11. Critically appraise and apply evidence even in the face of uncertainty and conflicting evidence to guide care tailored to the individual patient
12. Use pharmacogenomic testing when appropriate and/or describe its growing role in the management of diseases, including but not limited to:
    a. Hyperlipidemia
    b. Mood disorders
    c. Cancers
    d. Thrombus
    e. Human immunodeficiency virus

Medical Knowledge

Family medicine residents should demonstrate the ability to apply knowledge of the following:

1. Availability of genetic testing and/or treatment for conditions seen frequently in practice, such as:
    a. BRCA1/BRCA2 testing for hereditary breast and ovarian cancer
    b. Screening for colon cancer and implications of family history
    c. Newborn screening, including but not limited to:
        i. Congenital adrenal hyperplasia
        ii. Congenital hypothyroidism
        iii. Cystic fibrosis
        iv. Galactosemia
        v. Glucose-6-phosphate dehydrogenase deficiency
        vi. Glutaric acidemia type I
        vii. Hemoglobinopathy
        viii. Homocystinuria
        ix. Long-chain L-3-OH acyl-CoA dehydrogenase deficiency
        x. Maple syrup urine disease
        xi. Medium chain acyl-CoA dehydrogenase deficiency
        xii. Phenylketonuria
        xiii. Sickle cell disease
2. Basic medical knowledge of preconception/intrapartum counseling for carrier disease states (e.g., hemoglobinopathies, cystic fibrosis, spinal muscular atrophy)
3. Basic medical knowledge of intrapartum noninvasive prenatal testing (e.g., screening for trisomy/monosomy 13, 18, 21 and common sex chromosomal syndromes)
4. Understanding of NIPT as a screening test with additional invasive diagnostic testing (e.g., amniocentesis, chorionic villus sampling) needed in the event of a positive result
5. Risk scoring and focused testing for disease states (e.g., cancers that might or might not respond to treatments such as chemotherapy)
6. Screening for congenital cardiac cardiogenetic anomalies, such as hypertrophic cardiomyopathies, lipid genetics and coronary artery disease risk states, as well as long QT syndrome, if appropriate
7. Knowledge of psychotropic testing to analyze clinically important genetic variations that may impact how patients metabolize and/or respond to medications treating common mental health disorders

Interpersonal Communication

At the completion of residency, residents should be able to:

1. Perform a three-generation family history in assessing predisposition to disease
2. Effectively interview patients to obtain information about relevant genetic, environmental and behavioral risk factors
3. Provide unbiased information about the potential risks, benefits and limitations of genetic testing for informed consent for testing

4. Explain to patients the role of genetic factors in health maintenance and disease prevention
5. Counsel patients regarding the difference between clinical diagnosis of diseases and identification of genetic predisposition to disease for individuals, their families and the community
6. Recognize the influences of culture, related health beliefs and socioeconomic status in determining patient access to genetic information and services
7. Review the risks versus benefits of genetic approaches to treat disease (including pharmacogenomics and gene therapy)
8. Identify and accurately describe phenotypic features and/or anomalies using standardized nomenclature
9. Recognize complex syndromes or disorders
10. Promote a safe environment where patients and others involved in their care can actively engage in their care decisions
11. Assist patients and others involved in their care in locating reputable medical information on the internet and other sources
12. Discuss internet safety and protection of health information

Systems-Based Practice

At the completion of residency, residents should be able to:

1. Utilize pharmacogenomics to improve safety, efficiency and costs within the health care system
2. Recognize the importance of the genetics team as collaborators in the evaluation, diagnosis and decision to pursue or forgo treatment for patients undergoing genetic consultation
3. Seek consultation with medical genetics health care professionals
4. Refer patients to specialty treatment centers when appropriate
5. Advocate for patient care needs (e.g., community resources, patient assistance resources) with consideration for the limitations of each patient's payment model, including genetic testing through research

Practice-Based Learning

At the completion of residency, residents should be able to:

1. Critically appraise and apply evidence, even in the face of uncertainty and conflicting evidence, to guide care, tailored to the individual patient
2. Appropriately seek assistance from and refer to genetics experts
3. Appropriately make referrals for peer support resources, including but not limited to:
    a. Speech therapists
    b. Occupational therapists
    c. Dietitians
    d. Early intervention specialists

Professionalism

At the completion of residency, residents should be able to:

1. Recognize the philosophical, theological, cultural and bioethical perspectives influencing the use of genetic information and services
2. Appreciate 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
3. Recall the history of misuse of human genetic information (i.e., eugenics) and the obligation of health care professionals to "do no harm"
4. Safeguard the privacy and confidentiality of the genetic information of patients and warn of potential limitations
5. Demonstrate awareness of implicit bias, particularly in relationship to race and ethnicity

Implementation

Implementation of this curriculum should include longitudinal experience throughout residency training. Physicians with medical genetics expertise 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 inpatient and outpatient settings.

Resources

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