The University of Arizona Pediatric Residency Program

Primary Goals for Rotation

Genetics

1. **GOAL**: Understand the role of the pediatrician in preventing genetic disease, and in counseling and screening individuals at risk for these diseases.
2. **GOAL**: Differentiate disorders in patients associated with genetic predisposition or genetic disease from normal states or acquired disorders.
3. **GOAL**: Evaluate, treat, and/or refer patients with the presenting signs and symptoms that suggest a genetic disease process.
4. **GOAL**: Assist in diagnosis of genetic conditions and counseling of parents, under the supervision of a geneticist.
5. **GOAL**: Recognize and respond to urgent and/or severe conditions related to genetics and inherited metabolic disorders.
6. **GOAL**: Recognize genetic factors in common diseases of childhood and adulthood
7. **GOAL**: Demonstrate high standards of professional competence while working with patients under the care of a subspecialist. [For details see Pediatric Competencies.]

1. GOAL: Understand the role of the pediatrician in preventing genetic disease, and in counseling and screening individuals at risk for these diseases.

A. Provide routine genetic preventive counseling to all parents and patients that addresses:

1. Disorders identified in the neonatal screening program in one's state
2. Folic acid supplementation before and during pregnancy
3. Early and routine prenatal care and routine genetic screening for disease during pregnancy
4. Routine screening specific to certain ethnic groups
5. Avoidance of known teratogens during pregnancy (e.g. isotretinoin and alcohol), and reassurance about most substances that are not teratogenic

B. Provide prenatal and postnatal genetic preventive counseling to parents and patients with specific genetic conditions, addressing:

1. Genetic disorders with known or presumed inheritance patterns, based on a constructed pedigree
2. Expected course of known genetic disorders
3. Risk factors, including advanced maternal or paternal age and previous children with genetic conditions
4. Internet and other resources and support groups for known genetic disorders

C. Provide regular genetic screening:

1. Screen for known familial genetic disease processes using the appropriate method.
2. Describe screening methods, including CVS, amniocentesis, maternal serum screening and high-definition ultrasound in women over the age of 35 or at risk for having a child with a specific genetic problem
3. Identify screening programs to detect disease and carrier states in family members

2. GOAL: Differentiate disorders in patients associated with genetic predisposition or genetic disease from normal states or acquired disorders.

A. Describe general concepts that explain chromosome structure and spontaneous mutations, and molecular genetic techniques commonly used in diagnosis of genetic diseases.

B. Describe common patterns of Mendelian vs. non-Mendelian inheritance (autosomal dominant and recessive, X-linked, multifactorial, and the effect of maternal and paternal age), and demonstrate the ability to construct a pedigree.

C. Discuss unusual patterns of inheritance (mitochondrial defects, triplet repeat, imprinting).

D. Identify common diseases with known inheritance patterns and describe the mode of inheritance, including: cystic fibrosis, sickle cell anemia, Marfan syndrome, Huntington's Disease, neurofibromatosis, and familial cancer syndromes.

E. Identify common disorders with unusual inheritance patterns and describe the mode of inheritance, including: Fragile X, MERRF, and MELAS.

F. Explain the findings on clinical history and examination that suggest a known or potential genetic disorder or inborn error of metabolism.

G. Perform a thorough physical examination on a child suspected of a specific genetic disorder, identifying major and minor congenital anomalies that could be signs of an underlying genetic syndrome.

H. Describe how well child care differs in a child with a genetic condition, e.g., use of specific growth charts for specific conditions and physical findings.

I. Identify appropriate clinical and laboratory tests to help identify genetic diseases and inborn errors of metabolism. Explain the reason for the test to a family and interpret the results, with the assistance of a geneticist. The tests should include the following:

1. Chromosome analysis (both metaphase and prophase) and FISH testing for specific disorders
2. Plasma and urine amino acids, urine organic acids, ammonia level, venous pH, lactate, pyruvate, and blood acylcarnitine profile
3. Molecular testing for Fragile X
4. DNA mutational testing for selected disorders
5. Newer and future technologies developed for detection of genetic disorders (e.g., microarray technology)

J. Identify written and internet resources to aid in diagnosing a genetic or inborn error of metabolism, using physical findings along with laboratory examination.

K. Discuss the ethical, legal, financial and social issues involved in genetic testing of children for genetic disorders that may present in adulthood, testing children for carrier status, and providing medical care for patients with known fatal disorders.

L. Develop strategies to learn about future advances in the understanding of genetic disorders, in order to incorporate into one's practice improved screening, identification, counseling and management of such disorders.

M. Identify the indicators that would lead you to seek a genetics consult.

3. GOAL: Evaluate, treat, and/or refer patients with the presenting signs and symptoms that suggest a genetic disease process.

A. Create a strategy to determine if the following presenting signs and symptoms are caused by genetic disease or an inborn error of metabolism and determine if the patient needs treatment or referral.

1. Developmental delay
2. Dysmorphic features
3. Poor feeding
4. Vomiting
5. Failure to thrive
6. Seizures
7. Short stature
8. Hearing loss
9. Cleft lip/palate
10. Respiratory disorders
11. Obesity
12. Skin lesions

13. Hypotonia
14. Unusual behavior

4. GOAL: Assist in diagnosis of genetic conditions and counseling of parents, under the supervision of a geneticist.

A. Discuss the presenting signs and symptoms for commonly encountered genetic disorders (e.g., Trisomy 21, Turner Syndrome, Fragile X, neurofibromatosis, spina bifida, Marfan syndrome, achondroplasia) and identify accepted guidelines for care.

B. Develop a management plan for commonly encountered genetic disorders, identifying principles of long-term management, including use of disorder-specific growth charts and practice guidelines.

C. Provide primary care for and participate as a team member in medical and educational planning for a patient with a genetic disorder.

D. Identify resources in your community for diagnosis, genetic counseling, therapy, and psychosocial support of children with genetic defects and congenital anomalies.

5. GOAL: Recognize and respond to urgent and/or severe conditions related to genetics and inherited metabolic disorders.

A. Identify, explain, provide initial management and support, and seek urgent referral for the following genetic and/or metabolic conditions:

1. Infants presenting with symptoms that indicate the possibility of a severe inborn error of metabolism (e.g., metabolic acidosis, hyperammonemia, unexplained seizures, ketosis or hypoketosis, profound hypoglycemia)
2. Dysmorphic features found in chromosomal abnormalities that require prompt diagnosis in the perinatal period (e.g., Trisomy 13, 18, 21)
3. Unexplained critical illness or death suggestive of metabolic disorder, requiring collection of tissue samples before or at time of death
4. Developmental delay with signs or symptoms suggesting an underlying metabolic or genetic disorder
5. Physiologic changes or regression of milestones that suggest a possible metabolic etiology (e.g., urea cycle disorders, mitochondrial disorders, lysosomal storage diseases, abnormalities of organic/amino metabolism)

6. GOAL: Recognize genetic factors in common diseases of childhood and adulthood

A. Discuss current knowledge regarding the molecular basis of common childhood and adult conditions.

B. Identify the current and future uses of DNA testing in the office setting, including diagnosis of infectious diseases using DNA, pharmacogenetic testing for inborn errors of metabolic pathways prior to prescribing, DNA chips to identify genetic etiologies for complex disorders (e.g., congenital heart disease, seizure disorders, etc.).

C. Develop a strategy to incorporate concepts of molecular medicine into the everyday identification, treatment, counseling, and prevention of common disease processes.

7. GOAL: Demonstrate high standards of professional competence while working with patients under the care of a subspecialist.
A. **Competency 1:** Patient Care. Provide family-centered patient care that is development- and age-appropriate, compassionate, and effective for the treatment of health problems and the promotion of health.
1. Use a logical and appropriate clinical approach to the care of patients presenting for specialty care, applying principles of evidence-based decision-making and problem-solving.
2. Describe general indications for subspecialty procedures and interpret results for families.
B. **Competency 2:** Medical Knowledge. Understand the scope of established and evolving biomedical, clinical, epidemiological and social-behavioral knowledge needed by a pediatrician; demonstrate the ability to acquire, critically interpret and apply this knowledge in patient care.
1. Acquire, interpret and apply the knowledge appropriate for the generalist regarding the core content of this subspecialty area.
2. Critically evaluate current medical information and scientific evidence related to this subspecialty area and modify your knowledge base accordingly.
C. **Competency 3:** Interpersonal Skills and Communication. Demonstrate interpersonal and communication skills that result in information exchange and partnering with patients, their families and professional associates.
1. Provide effective patient education, including reassurance, for a condition(s) common to this subspecialty area.
2. Communicate effectively with primary care and other physicians, other health professionals, and health-related agencies to create and sustain information exchange and teamwork for patient care.
3. Maintain accurate, legible, timely and legally appropriate medical records, including referral forms and letters, for subspecialty patients in the outpatient and inpatient setting.
D. **Competency 4:** Practice-based Learning and Improvement. Demonstrate knowledge, skills and attitudes needed for continuous self-assessment, using scientific methods and evidence to investigate, evaluate, and improve one's patient care practice.
1. Identify standardized guidelines for diagnosis and treatment of conditions common to this subspecialty area and adapt them to the individual needs of specific patients.
2. Identify personal learning needs related to this subspecialty; systematically organize relevant information resources for future reference; and plan for continuing acquisition of knowledge and skills.
E. **Competency 5:** Professionalism. Demonstrate a commitment to carrying out professional responsibilities, adherence to ethical principles, and sensitivity to diversity.
1. Demonstrate personal accountability to the well-being of patients (e.g., following up on lab results, writing comprehensive notes, and seeking answers to patient care questions).
2. Demonstrate a commitment to carrying out professional responsibilities.

3. Adhere to ethical and legal principles, and be sensitive to diversity.

F. **Competency 6:** Systems-based Practice. Understand how to practice high-quality health care and advocate for patients within the context of the health care system.

1. Identify key aspects of health care systems as they apply to specialty care, including the referral process, and differentiate between consultation and referral.
2. Demonstrate sensitivity to the costs of clinical care in this subspecialty setting, and take steps to minimize costs without compromising quality
3. Recognize and advocate for families who need assistance to deal with systems complexities, such as the referral process, lack of insurance, multiple medication refills, multiple appointments with long transport times, or inconvenient hours of service.
4. Recognize one's limits and those of the system; take steps to avoid medical errors.

Procedures

A. GOAL: Technical and therapeutic procedures. Describe the following procedures, including how they work and when they should be used; competently perform those commonly used by the pediatrician in practice.

1. Wood's lamp examination of skin

B. GOAL: Diagnostic and screening procedures. Describe the following tests or procedures, including how they work and when they should be used; competently perform those commonly used by the pediatrician in practice.

1. Hearing screening
2. Radiologic interpretation: chest X-ray
3. Radiologic interpretation: CT of head
4. Radiologic interpretation: extremity X-ray
5. Radiologic interpretation: MRI of head
6. Vision screening

Adapted From

Kittredge, D., Baldwin, C. D., Bar-on, M. E., Beach, P. S., Trimm, R. F. (Eds.). (2004). APA Educational Guidelines for Pediatric Residency. Ambulatory Pediatric Association Website. Available online: www.ambpeds.org/egweb. [Accessed 03/26/2008]. Project to develop this website was funded by the Josiah Macy, Jr. Foundation 2002-2005.