

Pediatric Clinical Genetics
Course Directors
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Competency-Based Goals and Objectives:

The Department of Pediatric offers an elective in Clinical Genetics. At the completion of this rotation, Pediatric Resident will be expected to demonstrate competencies in six areas and will be prepared to incorporate competencies in the practice of pediatrics and other primary care specialties. Both Pediatric and Adult patients will be seen.

Competency 1: Patient Care: Provide family centered patient care that encompasses diagnosis and effective treatment of genetic disorders and give information promoting healthcare.

1. Effectively observe, communicate, and interact with patients, families, and other Healthcare workers to obtain histories, deal with difficult situations, and insure Proper record keeping.
2. Perform, record, present, and interpret appropriately focused physical exams
 - a. Pertinent to Pediatric Clinical Genetics.
Identify common dysmorphic features including
 1. Eye spacing
 2. Head circumference
 3. Ear size/shape
 4. Philtrum length
 5. Palate
 6. Neck
 7. Heart murmur
 8. Nipple spacing/location
 9. Genitalia
 10. Muscle tone
 11. Limb length
 12. Polydactyly
 13. Skin lesions
 - b. Identify infants presenting with symptoms that indicate the possibility of a severe inborn error of metabolism (e.g. metabolic acidosis, hyperammonemia, unexplained seizures) or chromosomal abnormalities that require prompt diagnosis in the perinatal period (e.g. Trisomy 13, 18, 21).
3. Integrate data obtained from the history, physical exam, and laboratory to construct a differential diagnosis for possible syndromes.
4. Provide primary care and participate as a team member in a medical and educational planning for patients with genetic disorders.
5. Participate in the process of genetic counseling and then review the experience with an experienced genetics counselor.

6. Differentiate genetic disorders from normal familial variation or non-genetic disorders.
 - a. Collect and document an appropriate family and personal history with emphasis on family history for genetic and prenatal history. Identify risks when present.
 - b. Perform appropriate oriented physical examination on proband and family with emphasis on identifying major and minor dysmorphic features which may be signs of underlying genetic syndromes.
 - c. Understand appropriate laboratory tests needed to support a diagnosis based on the history and physical.
 - d. Interpret test results.

Competency 2: Medical Knowledge Understand the scope of various genetic disorders integrating clinic features, routine laboratory values as well as specialized genetic tests in making a diagnosis

1. Recognize the importance of genetic diseases and congenital malformation in pediatrics.
 - a. Know the frequencies of genetic diseases and congenital anomalies at different stages, and their impact in morbidity and mortality.
 - b. Describe the impact on the family.
2. Appreciate the different types of genetic disorders and their diagnostic approach.
 - a. Explain the differences between etiology, pathogenesis, and phenotype; and identify the traditional and nontraditional etiologies.
 - b. Identify infants presenting with symptoms that indicate the possibility of a severe inborn error of metabolism (e.g. metabolic acidosis, hyperammonemia, unexplained seizures) or chromosomal abnormalities that require prompt diagnosis in the perinatal period (e.g. Trisomy 13, 18, 21).
 - c. Recognize unexplained critical illness or death suggestive of metabolic disorder, requiring collection of tissue samples before or at the time of death.
 - d. Identify developmental delay and other signs or symptoms suggesting an underlying metabolic or genetic disorder.
3. Participate in the diagnosis and management of common pediatric genetic diseases.
 - a. List presenting signs and symptoms and identify principles of long-term management of commonly encountered disorders (Down, Turner, Fragile X, neurofibromatosis, Fetal alcohol syndrome, Marfan syndrome, Facial clefting).
 - b. Identify signs and symptoms of congenital abnormalities diagnosed prenatally. Understand various methods of prenatal diagnosis used to confirm these diagnoses and follow-up.
 - c. Describe the disorders tested for in the State of Illinois Newborn screen. Identify the requirements used to determine what would be screened for on the newborn screen.
 - d. Explain the follow-up patients with a positive newborn hearing screen require.
4. Know the basic principles of genetic counseling:

- a. Identify and use resources in one's community for diagnosis, genetic counseling, therapy and psychosocial support of children with genetic defects and congenital anomalies and their families.
 - b. Discuss public health strategies to reduce risk for genetic disorders and congenital anomalies (e.g., early identification and screening programs to detect disease and carrier states, prenatal care, genetic counseling).
 - c. Interpret statistical analysis using Bayes analysis to determine recurrence risk.
 - d. Use information technology to access and manage clinical information and perform online searches about their patients and acquire knowledge of specific topics including testing.
 - e. Demonstrate commitment to collect appropriate screening histories, participate in neonatal screening programs, provide initial counseling and utilize resources for genetic counseling.
5. Pathology Studies
- a. Identify appropriate ordering of cytogenetic studies
 - b. Understand the indications for autopsy as well as the information which can be obtained through autopsy.
 - c. Identify the difference between clinical and laboratory diagnosis.

Competency 3: Communication and Interpersonal Skills Demonstrate interpersonal and communication skills in communicating the presence of a genetic disorder as well as the risk of recurrence within the family.

1. Elicit, record, and present focused history and physical examinations for common genetic disorders.
2. Communicate risk assessment and facilitate decision making in a nondirective manner.
3. Observe the health care provider obtaining informed consent including cost, legal, social and health issues.
4. Explain to physician and non-physician healthcare workers who consult the Genetics service as well as patients and families the findings of clinical exams and lab results. In addition to the risks, benefits and alternatives for possible therapies
5. Motivate and instruct patients and their families in promoting health care maintenance specific for the particular genetic disorder.
6. Demonstrate sensitivity in communicating with patients and families of how age culture, religion and personal beliefs and educational level can influence their perception of provider-patient interactions.
7. Observe families being given "bad news" with regards to a pregnancy or child.
8. Demonstrate ability in obtaining support information for the family on various disorders.
9. Demonstrate knowledge of the stages of grief and support resources available for families.

Competency 4 Problem Solving and Lifelong Learning: Demonstrate knowledge, skills and attitudes for continuous self-assessment, using scientific methods and evidence to investigate, evaluate, and improve one's patient care practice.

1. Identify and use resources to obtain up-to-date information dealing with genetic disorders and specific anticipatory guidance.
2. Search, evaluate, and present a critical review of scientific information in the medical literature on major topic relevant to a case or an area of knowledge and practice in which they are uncertain.

Competency 5: Professionalism Moral Reasoning and Ethics Demonstrate a commitment to carrying out professional responsibilities, adherence to ethical principles, and sensitivity to diversity.

1. Display behaviors that foster and reward the patients trust in the physician such as appropriate dress, grooming, punctuality, honest, courtesy respect for patient's confidentiality in the professional relationship with the patient.
2. Converse appropriately and behave with personal integrity in all elective activities and in interactions with peers, faculty, residents and non-physicians staff involved in the care and treatment of Clinical Genetic patients.
3. Work collaboratively as a member of a healthcare team including nurses, genetics counselors and laboratory personnel in providing comprehensive clinical genetic care.
4. Advocate the interests of patients over person interests while developing an appropriate balance between personal and professional beliefs and obligations.

Competency 6. System-Based Practice Understand how to practice quality health care and understand some of the legal issues involved in genetic testing.

1. Describe the impact of economic and health insurance issues on patients care with regards to testing for symptomatic and presymptomatic genetic disorders.
2. Interact with Social services and the home care team to facilitate patient access to nursing care and home care services for chronic genetic disorders outside of the hospital.
3. Advocate for patients with special needs such as Down syndrome.