

## GENETICS ROTATION

### Faculty:

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**Goal:** Develop competency in pediatric genetics.

**Competency Tools:** Multiple choice quiz, a review of a topic or literature, and presentation of topic/case in monthly genetics meeting.

### Learning Objectives:

1. Recognize the importance of genetic diseases and congenital malformation in pediatrics.
  - a. Identify 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. Understand how to 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 disorders, and prenatal history. Identify risks when present.
  - b. Perform appropriate oriented physical examination on proband and family with emphasis on identifying major and minor congenital anomalies which may be signs of underlying genetic syndromes.
3. 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 non-traditional etiologies.
  - b. Identify indications for testing in the primary care setting for genetic or metabolic disorders (e.g., for findings such as short stature, developmental delay, minor congenital anomalies, failure to thrive, seizures, family history suggestive of certain condition).
  - c. 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 neonatal period (e.g. Trisomy 13, 18, 21).
  - d. Recognize unexplained critical illness or death suggestive of metabolic disorder, requiring collection of tissue samples before or at the time of death.
  - e. Identify developmental delay and other signs or symptoms suggesting an underlying metabolic or genetic disorder.
4. 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).

5. Know the basic principles of genetic counseling:
  - a. Provide primary care and participate as a team member in a medical and educational planning for patients with genetic disorders.
  - b. Explain how to identify and use resources in one's community for diagnosis, genetic counseling, therapy and psychosocial support of children with genetic defects and congenital anomalies.
  - c. Participate in the process of genetic counseling and then review the experience with an experienced genetics counselor.
  - d. 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.)
  - e. Demonstrate commitment to collect appropriate screening histories, participate in neonatal screening programs, provide initial counseling and utilize resources for genetic counseling.

**Curricular Content:**

Curricular content is described in the subtext with the learning objectives.

**Skills Acquisition:**

1. Draw a pedigree.
2. Describe common methods of genetic diagnosis including genetic screening tests available, and identify resources for up to date information on this topic.
3. List the disorders screened for in Louisiana's neonatal screening program and provide initial counseling for an infant with a positive result.
4. Understand how to perform appropriate diagnostic searches using different books and computerized catalogs.
5. Perform a dysmorphologically oriented physical examination.
6. Become involved in the presentation or publication of unusual cases or new syndromes (time permitting).

**Reading Materials:**

To be determined by attending of the month.

**Rotation Requirements:**

1. Residents will participate in the genetics clinics at Charity and Children's Hospitals.
2. Residents will accompany the attending performing consultations.
3. Residents will participate with the genetic counselors as patients are counseled.
4. Residents will present a genetic topic at the end of the rotation.