GENETICS – DYSMORPHOLOGY: Case Based Approach

DATE: Monday, April 15 – 8:30am

LECTURER: Carolyn Jones, M.D., Ph.D.

KEY CONCEPTS AND LEARNING OBJECTIVES

1. Neurofibromatosis type I is a common disorder. There are several medical complications more common in patients with NF1
   a. Identify common clinical features seen in patients with NF1
   b. Describe complications of patients with NF1
   c. Understand the inheritance and frequency of NF1
   d. Recognize other disorders in the differential diagnosis for NF1

2. Connective tissue has many components. Abnormalities in any of the parts can result in instability of the connective tissue. Some body systems are more dependent upon the proper function of connective tissue than others. Marfan syndrome is one of the most common syndromes associated with connective tissue dysfunction
   a. Describe the common skeletal features.
   b. Understand which clinical studies are important in monitoring a patient with Marfan Syndrome?
   c. Understand the medical complications of this disorder.

3. There are many causes of hypotonia and muscle dysfunction. There are many genes involved in proper muscle function. Dystrophin is an essential component critical for proper muscle function. Mutations of the dystrophin gene can result in a varied disease depending upon the number of gene deleted.
   a. Compare the gene abnormality in Duchene’s Muscular Dystrophy (DMD) to Beckers Muscular Dystrophy (BMD). 
   b. Understand how are these disorders inherited?
   c. Describe the most common laboratory studies used to aid in the diagnosis.

4. Insulin is important in trafficking insulin into the cell. An increase number of insulin receptors can result in the cells growing faster. One disorder associated with overabundance of insulin receptors is Beckwith-Weideman Syndrome.
   a. Describe the features associated with Beckwith-Weideman syndrome
   b. Understand why it is important to diagnosis this disorder early
   c. Identify the complication that can result from this disorder.
d. **Understand anticipatory guidance for this disorder**

5. Failure to thrive, or poor growth in a child, can have many causes including nutritional, physical, single gene and teratogenic.
   a. **Recognize inherited and teratogenic causes for failure to thrive**
   b. **Understand the clinical and biochemical characteristics of Smith Lemli Opitz syndrome**
   c. **Understand the 4 major criteria needed to make a diagnosis of Fetal Alcohol Syndrome.**
   e. **Compare the frequency and symptoms of Fetal alcohol syndrome versus fetal alcohol affects.**
   f. **Identify the cause of failure to thrive in patients Prader Willi and Angelman syndrome**
   g. **Identify the genes and molecular causes in both Prader Willi and Angelman syndrome**