Inborn Errors of Metabolism

DATE: Monday, April 15, 2019 – 9:30 am

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KEY CONCEPTS AND LEARNING OBJECTIVES

1. Metabolic acidosis can result from many causes including diarrhea, renal tubular acidosis, diabetic ketoacidosis as well one of the inborn errors of metabolism,
   a. Understand the difference between metabolic acidosis with a normal anion gap and metabolic acidosis with an elevated anion gap.
   b. Recognize the laboratory values needed to access a possible inborn error of metabolism
   c. Understand differential diagnosis for an acidotic newborn.

2. There are many different disorders known as “Organic Academia’s”.
   a. Recognize common metabolic features of this group of disorders
   b. Identify common clues in some inborn errors including specific urine odors.
   c. Understand the importance of fluid management and diet for these patients.

3. Disorders of the urea cycle are seen when an individual is missing one of the enzymes in the Urea cycle. This cycle is important in disposing of nitrogenous waste.
   a. Describe the clinical characteristics of urea cycle disorders
   b. Identify the laboratory value abnormalities evaluated in considering this diagnosis.
   c. Understand the various inheritance patterns.

4. Fatty acid oxidation disorders represent a common group of inborn errors of metabolism. As a group, they occur when fat cannot be converted to energy. This process occurs in the mitochondria.
   a. Understand the process of β-oxidation.
   b. Describe the difference between carnitine and acylcarnitine.
   c. Recognized the symptoms and complications of fatty acid oxidation disorders, particularly MCAD.
   d. Understand the laboratory studies needed to make this diagnosis.
   e. Understand the treatment for affected patients.

5. Galactosemia is a metabolic disorder resulting from the inability to utilize galactose as an energy source. One of the most common sources of galactose is lactose.
6. Glycogen storage diseases are a group of disorders resulting from the inability to properly break down glycogen.
   a. Describe the 2 main systems most affected by this group of disorders
   b. Understand the common clinical features of these disorders.
   c. Understand the clinical features of McCordles disease and how it can be diagnosed.