Lysosomal Storage Disease Learning Objectives

DATE: Tuesday, April 16, 2019 – 8:30 am

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KEY CONCEPTS AND LEARNING OBJECTIVES

1. MPS type I is caused by the deficiency or absence of the enzyme α-1-Iduronidase. The clinical manifestations are progressive.
   a. Name the common clinical features of Hurler syndrome
   b. Describe the common skeletal features.
   c. Understand the difference between Hurler syndrome and Scheie.
   d. Identify and understand the mechanisms of action of various treatments.
   e. Recognize the inheritance pattern

2. Hunter syndrome or MPSII has many similar features to MPSI. It is caused by enzyme deficiency of iduronidase sulfatase deficiency.
   a. Describe the features of Hurler syndrome that differentiate it from Hurler syndrome
   b. Describe complications seen in Hunter syndrome
   c. Understand the inheritance pattern.

3. Fabry Disease is an X-linked recessive lysosomal storage disorder. It results in early renal failure in males. While the gene is located on the X-chromosome, females can have symptoms of this disorder similar to males.
   a. Compare the clinical features of this disorder in males and females
   b. Described the classic clinical features in male
   c. Understand treatment options for affected patients,

4. Gaucher disease is the most common lysosomal storage disorder. It is inherited in an autosomal recessive fashion. Symptoms often do appear until adulthood.
   a. Describe the features associated with Gaucher Disease
   b. Identify the complication that can result from this disorder.
   c. Understand anticipatory guidance for this disorder
   d. Identify the various treatment options.

5. Infantile Pompe’s disease is characterized by early onset cardiomyopathy and death. It is caused by the deficiency of the enzyme alpha galactosidase A.
a. Understand the clinical course for this disease without treatment.
b. Understand the body systems most affected by the absence of this enzyme.
c. Identify treatment options.
d. Recognize the various features of adult onset disease.

6. Tay Sachs disease is a rare lysosomal storage disease affecting primarily the brain. While it was seen more commonly in individuals of Ashkenazi Jewish descent, the majority of babies diagnosed with Tay Sachs are not of Ashkenazi descent.
   a. Recognize the unique clinical features seen in Tay Sachs disease including cherry red spot.
   b. Understand the inheritance pattern.