Hurler Syndrome

Hurler’s syndrome is a mucopolysaccharidosis lysosomal storage disease caused by a deficiency in the enzyme alpha L iduronidase. Without this enzyme, cells are unable to break down carbohydrates which results in the accumulation of glycosaminoglycans including dermatan sulfate and heparan sulfate. Accumulation of these substrates result in developmental delay and specific physical findings including gargoylism. Patients also suffer from airway obstruction and hepatosplenomegaly. This disease is inherited in an autosomal recessive pattern.

PLAY PICMONIC

Pathophysiology

Autosomal Recessive
Recessive-chocolate
Hurler syndrome is inherited in an autosomal recessive modality. This means that two copies of the abnormal gene (one from each parent) must be present in order for this disease to develop in a patient.

Mucopolysaccharidoses
Mucous-polly-sack
Mucopolysaccharidoses are a set of diseases marked by the inability of the body to breakdown long chain sugar carbohydrates which results in the buildup of these long chain substrates. After a period of time and significant substrate buildup, they can deposit in a variety of tissues to cause disease.

Alpha-L-Iduronidase Deficiency
Afro-Iditarod-race-dog Broken
This is the enzyme deficient in Hurler syndrome which results in the accumulation of dermatan sulfate and heparan sulfate.

Increased Heparan Sulfate
Up-arrow Hippie-run with Sulfur-match
This is a glycosaminoglycan which accumulates in mucopolysaccharidoses including Hunter and Hurler syndromes.

Increased Dermatan Sulfate
Up-arrow Deer-man-tan with Sulfur-match
This is a glycosaminoglycan which accumulates in mucopolysaccharidoses including Hunter and Hurler syndrome.

Signs and Symptoms

Hepatosplenomegaly
Liver-and-spleen-balloons
Hepatosplenomegaly in Hurler syndrome is enlargement of the liver and spleen due to accumulation of dermatan sulfate and heparan sulfate in the liver and spleen.

Developmental Delay
Developmental-bus
Patients with Hurler syndrome suffer from developmental delay due to substrate buildup which affects brain development.

Gargoylism
Gargoyle
Gargoylism is a term used to describe prominent facial features in patients with Hurler syndrome. Some features include a flat face, depressed nasal bridge, and bulging forehead.

Corneal Clouding

Corn Clouds

Patients will present with blurred vision and opacification on ophthalmologic exam due to deposition of accumulated substrate in the cornea.

Airway Obstruction

Obstructed Airway

Airway obstruction is a frequent concern of patients with Hurler syndrome. This is often secondary to glycosaminoglycan deposition in the soft tissues of the neck, cervical abnormalities, as well as craniofacial abnormalities which can cause obstruction.