Pompe Disease

Pompe’s disease is type II glycogen storage disease cause by defect in lysosomal alpha-1,4-glucosidase enzyme. This enzyme is essential for the lysosomal degradation of glycogen to glucose. The enzyme is particularly important within the heart. Therefore, Pompe’s disease is particularly noxious to the heart. Patients also present with hepatomegaly due to build up of glycogen in the liver and hypotonia. However, this reaction is not essential for standard blood glucose maintenance, and patients with Pompe’s disease will have normal blood glucose levels.



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Pathophysiology

Type II Glycogen Storage Disease
(2) Tutu beside Glider
Pompe's disease is a type II glycogen storage disease.

Autosomal Recessive
Recessive-chocolate
Pompe disease is inherited in an autosomal recessive modality. This means two copies of the abnormal gene must be inherited in order for the disease to develop.

Deficiency of Lysosomal Alpha 1,4 Glucosidase
Broken Lysol-can with an Afro using a (1) Wand and (4) Fork with Glue-daisies
This enzyme hydrolyzes terminal 1-4 linked alpha-glucose residues to release a single alpha-glucose molecule from glycogen.

Acid Maltase
Acidic-lemon Maltese
Acid maltase is an alternative name for alpha 1-4 glucosidase that cleaves maltose residues, a nearly functional equivalent to alpha glucose.

Signs and Symptoms

Hypertrophic Cardiomyopathy
Hiker-trophy and Heart-mayo-partyhat
Enlargement of the heart occurs due to alterations in normal metabolic activity, resulting in increased strain and cardiac muscle hypertrophy. Cardiomyopathy and heart failure are the most common cause of death in patients with early-onset Pompe disease.

Hepatomegaly
Liver-balloon
Enlargement of the liver results from increased glycogen storage with limited breakdown and mobilization ability. The liver is particularly affected, because the glycogen is heavily stored in the liver.

Macroglossia
Macaroni-glass tongue
An early finding in those with Pompe disease is macroglossia, described as a markedly enlarged tongue.

Hypotonia
Floppy Hippo-baby
Within the first few months of life, the infantile form of Pompe disease presents with marked hypotonia. This is described as decreased muscular tone throughout the body. In adult Pompe disease, this is often a late manifestation, and patients become hypotonic after developing proximal muscle weakness,

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mimicking what is seen in limb-girdle muscular dystrophy.

Diagnosis

Increased Glycogen within Lysosomes

Up-arrow Gliders with Lysol

Pompe disease is the only glycogen storage disease which causes lysosomal metabolism defects. Alpha glucosidase is a lysosomal enzyme, and this enzyme defect results in excessive glycogen storage and accumulation within lysosomes. This leads to cellular injury and interruption in other organelle functioning.

Increased Lactate Dehydrogenase (LDH)

Up-arrow Lake-dehydrator

Lysosomal glycogen accumulation and subsequent cellular dysfunction in cardiac, smooth and skeletal muscle, as well as many other tissues, results in elevated lactate dehydrogenase (LDH) levels, among other lab values indicating abnormally high rates of cellular damage.

Increased Creatine Kinase (CK/CPK)

Up-arrow Calvin Klein model

Due to cardiac myopathy and muscular involvement, patients display increased creatine kinase levels in their bloodstream. Creatine kinase is an enzyme found in skeletal muscles and the heart, and elevated levels are seen in heart attacks, when the heart muscle is damaged, or in conditions that produce damage to the skeletal muscles.

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