Cori Disease

Cori's disease is a type III glycogen storage disease, and it results from a deficiency in the debranching enzyme alpha-1,6-glucosidase. The cells are able to breakdown glycogen and mobilize it into glucose, but there is a limitation in breaking down the branched bonds. This ultimately results in some glycogen build up due to the body's inability to break down the alpha 1, 6 bond between glucose residues. The clinical presentation is similar to type 1, Von Gierke's, but less severe. Patients still suffer from hypoglycemia, but not to the extent as seen in type 1. Additionally, patients often have stunted growth due to abnormal glycogen metabolism.

PLAY PICMONIC

Pathophysiology

Type III Glycogen Storage Disease

(3) Tree with Gliders

Cori's disease is also referred to as type III glycogen storage disease. This is an autosomal recessive disorder, where patients are unable to convert branched glycogen polymers to glucose due to a deficiency of debranching enzyme.

Deficiency of Alpha 1, 6 Glucosidase

Broken Afro-guy with (1) Wand and (6) Mask Stuck in Glue-daisies

Alpha-1,6 glucosidase or glucosidase is a component of debranching enzyme, and breaks the alpha 1, 6 bond in glycogen. Thus, this enzyme helps mobilize glucose.

Debranching Enzyme

Enzyme Cutting-branches

Branching bonds of glycogen are essential for effective storage. In Cori's disease, these bonds cannot be broken due to a deficiency in the debranching enzyme.

Limit-Dextrin Accumulation (In Cytosolic Structures)

Limit-Desks Accumulating at Side-toe-sail Boat

In this disorder, patients have an inability to convert branched glycogen polymers to glucose. Because limit dextrin is a remaining polymer produced after hydrolysis of glycogen, patients develop limit dextrin accumulation in their cytosol.

Signs and Symptoms

Hypoglycemia

Hippo-glue-bottle

While there is glycogenolysis ability in Cori's disease, it is limited due to the inability to breakdown branched glycogen molecules. This limitation in the body's ability to acutely mobilize glucose stores can result in transient hypoglycemia.

Muscle Weakness

Droopy Muscle

Patients have abnormal glycogen accumulation in muscle tissues in Cori disease. The accumulated glycogen is structurally abnormal and impairs the function of muscles, leading to weakness. This side effect of Cori disease is especially pronounced in adult form.

Hypotonia

Floppy Hippo-baby

Patients have abnormal glycogen accumulation in Cori disease. This is structurally abnormal and impairs the function of muscles. Often, the first sign and of disease in infants is poor muscle tone, or hypotonia.

Stunted Growth

C-clamp Stunting Growth

Growth is limited because the body cannot mobilize glycogen sufficiently and sustain adequate glucose levels for the body.

Hepatomegaly

Liver-balloon

Due to the deficiency of the debranching enzyme, there is accumulation of residual glycogen in the liver, which leads to hepatomegaly.

Diagnosis

Normal Blood Lactic Acid Levels

Normal-bell-curve at Lake Acidic-lemon

The cells are still able to undergo gluconeogenesis and create free glucose residues, so lactic acid levels are within normal limits.

Gluconeogenesis Intact

Glue-genie

Unlike in Von Gierke disease, in Cori disease gluconeogenesis is intact. This means that patients are able to generate glucose from non-carbohydrate substances like lactate, glycerol, and glucogenic amino acids.

Increased LFT's

Up-arrow Liver with Test-tubes

Patients with Cori disease can develop hepatomegaly from abnormal amounts of accumulating glycogen. If the liver is involved, patients can display elevations in liver function enzymes (LFTs) on lab testing.

Increased Creatine Kinase (CK/CPK)

Up-arrow Calvin Klein Model

The abnormal glycogen accumulation in Cori disease is structurally abnormal and impairs the function of muscles. Additionally, in times of glucose need, muscle protein is metabolized as a glucose source. These factors lead to lab findings of increased creatine kinase, which is a marker of muscle breakdown.

Treatment

Continuous Feeding

Infinite Feeding

It is recommended to provide frequent daytime feedings, along with continuous nasogastric tube feedings at night to infants up until the age of 3 years old. This is done to ensure they maintain satisfactory blood glucose levels.

High Protein Diet

Protein Filled Meat

Because gluconeogenesis is intact and protein can be used as a source of glucose, patients are recommended to have a high protein diet. A high protein diet prevents breakdown of muscle protein in times of glucose need, preserving muscles.

Cornstarch (Uncooked)

Corn-star

Uncooked cornstarch is a recommended dietary supplement in patients because it is a slow-release form of glucose.