Von Gierke Disease

Von Gierke’s disease is a type I glycogen storage disease cause by a deficiency in glucose-6-phosphatase. This key enzyme is used for breaking down glycogen to make glucose when blood glucose levels are low. Without this enzyme, the body is unable to manufacture the glucose needed to maintain blood sugar levels. Therefore, patients suffer from severe fasting hypoglycemia and develop elevated blood lactate levels as they use alternative fuel sources. Patient’s also develop severe hepatomegaly and enlarged kidneys due to the accumulation of glycogen in these organs.



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Pathophysiology

Autosomal Recessive
Recessive-chocolate
Von Gierke 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.

Type I Glycogen Storage Disease
#1 Glider jerky
Von Gierke's disease is a type I glycogen storage disease.

Glucose-6-Phosphatase Deficiency
Glue 6 Phosphate-P is Broken
This is the enzyme that is essential for the final step of glycogenolysis. If deficient, the patient is unable to mobilize glucose stores via glycogen breakdown.

Signs and Symptoms

Increased Glycogen in Liver
Up-arrow Glider in Liver
Without the glucose 6 phosphatase enzyme participating in glycogen breakdown, glycogen stores are unable to be mobilized and accumulate in the liver. It is important to note that patients can make glycogen but cannot break it down.

Hepatomegaly
Liver-balloon
Hepatomegaly, or an enlarged liver, occurs due to the abundance of glycogen accumulating in the liver.

Enlarged Kidneys
Kidney-balloon
Glycogen is also stored in the kidneys, thus it also accumulates in the kidney, much like it does in the liver.

Severe Fasting Hypoglycemia
Extremely Skinny Hippo-glue-bottle
Patients lack the ability to maintain blood sugar, because when blood sugar levels are low, glycogen cannot be effectively broken down to glucose in the liver.

Increased Triglycerides
Up-arrow Triceratops
Because there is a deficiency of glucose-6-phosphatase, glucose-6-phosphate molecules are shunted into other metabolic pathways. These glucose-6-phosphate molecules are converted into triglycerides, which are exported for storage in adipose tissue as fat.

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Increased Uric Acid (Gout)

Up-arrow Unicorn with Acidic-lemon

In Von Gierke disease, increased uric acid accumulates because of numerous reasons. A concurrent lactic acidosis in this disorder leads to decreased urinary excretion of uric acid. Additionally, because glucose-6-phosphatase is deficient, glucose-6-phosphate is shunted through the pentose phosphate pathway to create uric acid.

Increased Lactic Acid

Up-arrow Lake Acidic-lemon

Lactate increases as the body is forced to utilize alternative means for fuel when blood glucose levels are insufficient.

Doll-like Facies

Doll Face

Patients with Von Gierke disease have a characteristic rounded "doll-like facies" due to fat deposition.

Treatment

Continuous Oral Glucose (or Cornstarch)

Infinite Oral Glue

Because patients are hypoglycemic, and cannot participate in glycogenolysis, they must continuously ingest oral glucose. Cornstarch is another dietary option, as it provides gradually digested glucose, and decreases the necessity for constant feeds.

Avoid Fructose and Galactose

Avoid Fruit-toast and Galactic-toast

Fructose and galactose should be avoided in Von Gierke patients, as these sugars must first be converted to glucose-6-phosphate before they can be utilized in the body.

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