Andersen Disease (Type IV Glycogen Storage Disease)

Andersen disease is characterized by deficient glycogen branching enzyme activity caused by an autosomal recessive mutation in the GBE1 gene. Clinical manifestations are seen in early infancy and include hepatosplenomegaly, failure to thrive, cirrhosis, muscular weakness, hypotonia, cardiomyopathy, and hypoglycemia. Neuromuscular form of the disease can appear at any age. Management consists of supportive care.

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Pathophysiology

Branching Enzyme
Branch Enzyme
Andersen disease is characterized by a lack of glycogen branching enzyme activity, which results in an accumulation of abnormal glycogen in body tissues such as the liver and muscle.

GBE1 Mutation
GB-Electronic-1 Mutant
The mutation occurs in the GBE1 gene, which is located on chromosome 3p14. As a result, an abnormal glycogen structure known as polyglucosan accumulates.

Autosomal Recessive
Recessive-chocolate
Andersen disease is inherited in an autosomal recessive manner. This mode of inheritance is commonly seen in enzyme deficiencies.

Sign and Symptoms

Hepatosplenomegaly
Liver-and-spleen-balloons
Because the liver and muscle contain the most glycogen, they are the organs most affected by Andersen disease. Hepatosplenomegaly is caused by glycogen accumulation in the liver and spleen.

Failure to Thrive
Very Skinny Baby
Failure to thrive, along with hepatosplenomegaly, is the first symptom that appears in Andersen disease patients during their first months of life. It is defined as the patient's inability to grow and gain weight.

Cirrhosis
C-roses-on-liver
The abnormal glycogen in the liver causes cellular injury and organ dysfunction by acting as a foreign body. As a result, patients develop cirrhosis.

Muscular Weakness
Weak-drooping-muscle
Excess insoluble glycogen in the skeletal muscle of Andersen disease patients causes muscular weakness, exercise intolerance, and muscle atrophy.

Hypotonia
Floppy Hippo-baby
Severe hypotonia, characterized by very low muscle tone, is another manifestation of Andersen disease.

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Cardiomyopathy

Heart-mayo with Party-hat

Excess insoluble glycogen in the heart of Andersen disease patients will lead to cardiomyopathy.

Hypoglycemia

Hippo-glue-bottle

Hypoglycemia does not appear until the condition has progressed. Cirrhosis is commonly associated with it.

Management

Supportive Care

Supportive IV bags

Treatment of Andersen disease includes supportive care.

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