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HMB 401 ASSIGNMENT

QUESTION:

Discuss in details the inborn error of metabolism of Cori disease or Type III glycogen storage disease.

ANSWER:

In GSD III deficiency of the glycogen debrancher enzyme results in massively accumulated glycogen resembling glycogen with a shorter outer chain which is harmful for hepatocytes. The liver and muscle affecting form GSD IIIa accounts for 80% of all cases whereas GSD IIIb involves solely the liver. Glycogen debrancher enzyme has two independent catalytic enzymes and selective loss of only one of the two debranching activities results in GSD IIIc (glucosidase deficiency) and GSD IIId (transferase deficiency) which are very rare.

Disorders of glycogen metabolism may affect the liver or muscles or even both. Hepatomegaly and hypoglycaemia are the main findings in liver-affecting GSD. Laboratory investigations may show lactic acidosis, elevated transaminases, hyperlipidaemia, hypothyroidism, prolonged bleeding time, iron refractory anaemia and hyperuricaemia with considerable heterogeneity within the different types.

Most patients with GSD III show liver as well as muscle involvement (GSD IIIa). In about 15% of cases the disease is limited to the liver (GSD IIIb). Hepatomegaly, hyperlipidaemia and growth retardation will improve with age and may even disappear after puberty. Tendency to hypoglycaemia is much less than in GSD I.
Liver fibrosis is seen early in the disease process. However, hepatic involvement is considered to be self-limited usually without any symptoms of hypoglycaemia or active liver disease after the second decade of life. Onset of overt cirrhosis is atypical in the vast majority and liver cirrhosis or HCC has been reported only in few cases.
In GSD III hepatomegaly, hypoglycaemia, hyperlipidaemia, and growth retardation improve with age and disappear after puberty. However, liver cirrhosis and/or hepatocellular carcinoma may occur. Muscle weakness can become prominent in adults.