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Type III Glycogen Storage Disease (Cori's Disease) – A Case Report

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Abstract:
Glycogen storage disease type III is an metabolic disease caused due to deficiency of debrancher enzyme[2] which is necessary for cleaving the 1,6 linkage of outer branches of glycogen molecule, resulting in accumulation of glycogen with short branch points or limit dextrins.[3]
In this disease the patients mainly presents with hepatomegaly, hypoglycemia, poor growth, hyperlipidemia, elevated liver enzymes.[2]
Here we discuss a case of GSD type III with predominantly liver involvement.

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I. Case Report:

A 10 months old male child was admitted with complaint of gradual distension of abdomen and floppiness of all four limbs for last 4 months associated with generalised tonic clonic seizure for last 3 days.

There was history of third degree consanguineous marriage of parents. History of 2 siblings death at infancy is present.

On admission the patient was found to be drowsy, hypoglycemic with chubby cheeks and protruberent abdomen. On examination, there was hepatomegaly (liver span was 15 cm), gross hypotonia in all four limbs. Other system examinations were normal.

His laboratory findings revealed

- Mild normocytic normochromic anaemia
- Hyperlipidemia (serum cholesterol 252 mg/dl, serum triglyceride 693 mg/dl)
- Serum uric acid 3.7 mg/dl
- Serum lactic acid 2 mmol/L
- LFT- WNL
- MRI brain – WNL
- EEG – normal study
- CSF study – WNL
- USG W/A – liver 15 cm with moderate fatty change
- ECHO- trivial TR, good bi ventricular function

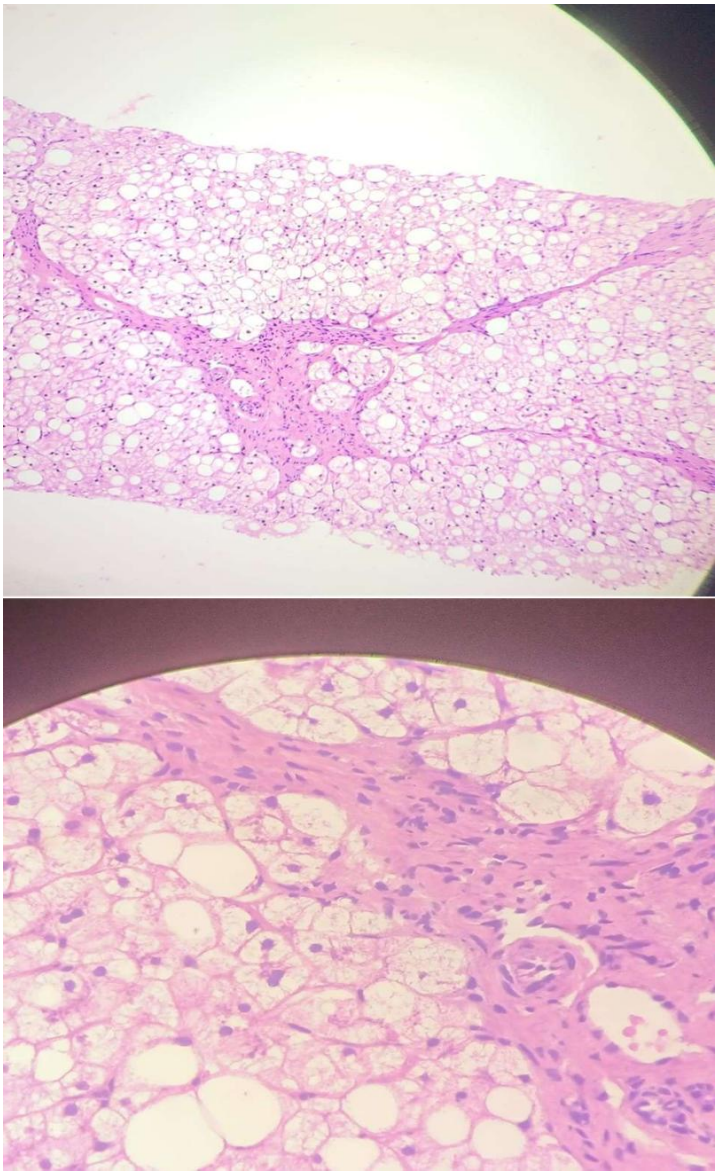
Based on the findings a provisional diagnosis of glycogen storage disorder was suspected and a liver biopsy was performed and report was awaited.

Meanwhile the patient was treated with IV dextrose, glucose infusion for hypoglycemia. The patient became convulsion free, started with small frequent oral feeds. No further hypoglycemic episode was noted. liver biopsy report came after 4 days.

Liver biopsy:

- Sections of liver show distortion of architechture due to broad fibrous bridges forming micronodules.
- The portal tracts show portal , portoseptal and occasional portoportal bridging fibrosis.
- The hepatocytes are diffusely swollen with pyknotic and eccentrically located nuclei and rarefied cytoplasm containing wisps of pinkish material and glycogen.
- The hepatocytes are stained bright magenta with the PAS stain and diastase sensitivity

Impression **– *Liver biopsy consistent with Glycogenosis type III with micronodular cirhhosis.***



I I . Discussion:

Glycogen storage disorder generally presents in early childhood and has an incidence of 1:50000 to 1:70000. Hepatic glycogenoses cause mainly hepatomegaly and fasting hypoglycemia whereas the muscle glycogenosis cause muscle weakness and cramps during exercise.

GSD type III is an autosomal recessive disorder with deficiency of debranching enzyme.[3] Approximately 85% patients[4] are GDE deficient in liver and muscle (type IIIa) and 15% patients have GDE absent in liver but retained in muscle (type IIIb) .[4]

In this case hepatic involvement is predominant with mild skeletal involvement. There is protruded abdomen due to hepatomegaly with increased serum cholesterol and doll like face.

The patients have fasting hypoglycemia but unlike patients of GSD type I , infants with GSD type III can tolerate longer fast due to active phosphorylase and intact gluconeogenesis.[5] So only when the feeding time is few hours apart or the child is unable to take feed , hypoglycemia can occur. The differentiation between type I and type III GSD was made by the intravenous glucagon test in the immediate post prandial phase when there is usually a rise in blood sugar level in type III but not in type I.[5]

Another differentiating point between type I and type III GSD is that no hyperuricemia or lactic acidosis is seen in type III whereas it can be seen in type I .[3] Renal involvement is also seen in type I but not in type III.[2]

In liver histology, the presence of fibrous septa and paucity of fat is a notable distinction from type I Glycogenosis.[5]

GSD type III can be managed by frequent high protein feedings.[1]Frequent day time meals with

complex carbohydrate and proteins can prevent hypoglycemia. In case of infants, gastric drip feeding may be introduced.

In our case patient was advised to take frequent small meals with cornstarch supplements[1] and advised to avoid simple sugar to prevent sudden spike of blood glucose and to take frequent meals at night time also.

Follow up plan is to measure blood glucose, ketones, LFT, CK, lipid profile, growth monitoring , ECG and Echocardiogram to monitor cardiomyopathy, also to monitor the liver fibrosis progressing to Hepatocellular carcinoma.

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