A Rare Case of Glycogen Storage Disease Presenting as Liver Failure in an Adult

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Background

- Glycogen storage disease (GSD) is an autosomal recessive disease, affecting 2.3 in 100,000 children in the US,
- GSD (III) is caused by abnormalities in AGL gene, causing deficiency of the glycogen debranching enzyme resulting in the accumulation of glycogen in tissues.
- It usually presents in early childhood with hypoglycemia and hepatomegaly.

Case Presentation

- This patient was a 22-year-old previously healthy man with a one-month history of jaundice, fatigue, weight loss, and pain in the abdomen with no past history of medical or surgical illness

Vitals & Physical Examination

- At presentation: vitals were stable, alert, pale, jaundice +, bruises + on his upper and lower limbs, full cheeks and thin limbs
- P/A- Palpable splenic tip, no hepatomegaly or ascites
- Musculo skeletal system examination – Power- 3/5 in all muscle groups, more pronounced in hips, shoulder
- CNS examination- weak DTRs in knee and ankle

Laboratory Investigations

WBC	**4X10*3/uL(4.5-11 K/uL)**
Hemoglobin	8.6 g/dL (12-16 g/dL)
Platelet count	100,000/uL
LFT(Bilirubin/AST/ALT)	92umol/L, 30U/L, 48U/L
Serum Albumin	23 g/L
PT/INR	20sec /1.68
Random Blood Sugar	5.0 umol/L
Lipid profile	normal
Autoantibody screens	Negative ANA, anti LK1, anti SMA

Clinical Course

USG abdomen ruled out ascites, hepatomegaly and dilated portal vein.

- He had, normal serum copper, ferritin and alpha fetoprotein.
- **Bone marrow biopsy** showed decreased cellularity
- **Liver biopsy** revealed significant glycogen load in hepatocytes on special stains along with periportal septal fibrosis suggestive of GSD with cirrhosis .
- **Muscle biopsy** showed vacuolar myopathic changes with focal endomysial fibrosis and inflammation, Vacuoles were filled with glycogen and debranching enzyme level was 18%
- **Echocardiography:** No cardiomegaly and normal ejection fraction
- He was advised for high protein and low complex carbohydrates diet with frequent feeds avoidance of simple sugars and fasting.
- He was advised regular follow ups to evaluate liver function, cardiac functions and myopathy

Discussion

- Glycogen storage disease type III presents usually in infancy with hypoglycemia and hepatomegaly
- It is associated with full cheeks, hypertriglyceridemia, hypoglycemia, immunodeficiency, intellectual disability, short stature, and myopathy
- GSD IIIa is present in 85% of cases; and presents with hepatic and skeletal muscle involvement.
- GSD IIIb, is found in the15% and involves only liver
- It is diagnosed by histology and genetic testing and the measurement of debranching enzyme levels in liver, heart or skeletal muscles
- In adults' liver involvement is less prominent but eventually most develop cardiac involvement and myopathy.
- It can cause cirrhosis with liver failure and lesions can undergo malignant transformations. Treatment aims to prevent hypoglycemia through carbohydrate meals and night feeds with cornstarch.
- Prognosis is determined by muscular and cardiac symptoms as well as liver fibrosis/cirrhosis

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