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Letter to the Editor

Budd-Chiari Syndrome in Gaucher Disease Type III in an Adult Libyan Male: Letter to the Editor

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To the editor,

Gaucher Disease (GD) is the most common lysosomal storage disorder. The prevalence of GD is approximately 1/100,000, and type III GD accounts for 5% of cases. [1] It is an autosomal recessive disease due to a GBA gene mutation, leading to glucocerebrosidase enzyme deficiency. [1,2] Gaucher disease (GD) is categorized into three types according to clinical presentation: [3] Type I, which is non-neuronopathic and most common, particularly among Ashkenazi Jews; Type II, which is acute neuronopathic and marked by significant neurological involvement and high mortality rates; and Type III, which is subacute neuronopathic, exhibiting both systemic and neurological symptoms. In this report, we discuss a 24-year-old man from Libya diagnosed with GD type III. His diagnosis was established at the age of one due to symptoms including pallor, poor appetite, and hepatosplenomegaly. Laboratory tests indicated a hemoglobin level of 5.6 g/dL, chitotriosidase activity of 18,742 $\mu\text{mol/L}$, and an angiotensin-converting enzyme level of 251 U/L. Genetic analysis confirmed a homozygous L444P mutation. He underwent splenectomy at the age of three, and enzyme replacement therapy (ERT) was administered intermittently with regular follow-ups until 2011. In December 2023, the patient experienced two weeks of abdominal pain, distension, and fatigue. A physical examination revealed ascites, dilated abdominal veins, and an enlarged liver and spleen. Laboratory findings are detailed in Table 1.

Budd-Chiari syndrome was suspected when abdominal ultrasound showed severe liver parenchymal disease, enlarged caudate lobe, and portal vein thrombosis. The initial treatment regimen consisted of furosemide, spironolactone, lactulose, and rivaroxaban. However, anticoagulation therapy was discontinued due to prolonged international normalized ratio (INR). The patient required multiple paracentesis procedures for ascites, which led to an improvement in the symptoms. However, the patient developed recurrent hyponatremia and ultimately passed away three months later.

Neurological symptoms in GD type III typically manifest during childhood or adolescence and include progressive myoclonic epilepsy, cerebellar ataxia, and spasticity. Liver and huge spleen enlargement are characteristic abdominal findings. Pancytopenia, which can manifest as anemia, thrombocytopenia, and skeletal abnormalities, is another presentation.

Hepatic vein outflow obstruction is a rare complication in GD, presenting with ascites, hepatomegaly, and abdominal pain. [4] GD's hepatic vein outflow obstruction occurs due to thrombosis or external compression. [5] GD treatment primarily involves enzyme replacement therapy (ERT), which alleviates systemic symptoms; however, it has limited efficacy on the disease's neurological progression. [6] Management includes anticoagulation, thrombolysis, or surgical interventions such as transjugular intrahepatic portosystemic

shunting. Budd-Chiari syndrome is very rare in GD patients and significantly impacts the patient's prognosis. Massive splenomegaly produces pain, compression, and thrombosis. Splenectomy worsens hypercoagulability and recurrent infection, thus, it is advisable to avoid this procedure. Our patient underwent ERT that reduced systemic GD disease symptoms but did not prevent or improve his neurological and thrombotic complications. Treatment of Budd-Chiari syndrome requires anticoagulation and symptomatic treatment.

Table 1. Summary of laboratory results at the presentation of Budd-Chiari syndrome.

Investigation	Results
Liver function	alkaline phosphatase (575 IU/L), alanine transaminase (71 IU/L), and aspartate aminotransferase (49 IU/L).
Coagulation profile	INR of 1.1.
Renal function	Renal function and potassium were normal, and the serum sodium was (109 mEq/L)
Ascitic fluid analysis	Protein concentration (5.5 g/dL) and the SAAG >1.1

Serum-ascites albumin gradient (SAAG).

In conclusion, the overlap between Gaucher disease type III and Budd-Chiari syndrome is rare, which makes it difficult to diagnose and treat. Effective treatment and early detection of problems improve outcomes. The increased risk of coagulopathy in Gaucher disease, along with the presence of hepatic venous outflow obstruction, hampers the prognosis.

PATIENT CONSENT

A written informed consent was obtained from the patient for publication of this case report.

AUTHORS' CONTRIBUTION

All authors contributed to the completion of this work. The final manuscript was read and approved by all authors.

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CONFLICT OF INTEREST

None.

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