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The Intersection of Rare: Gauche Disease Complicated by Cirrhosis, Hemochromatosis, and Acute Coronary Syndrome

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BACKGROUND & AIMS

Gaucher disease, a rare lysosomal storage disorder, is known for its diverse clinical manifestations primarily affecting the reticuloendothelial system. However, its association with cirrhosis, hemochromatosis, and acute coronary syndrome is exceedingly rare and scarcely reported in the literature.

This abstract aims to present a rare case report of Gaucher disease manifesting with cirrhosis, secondary hemochromatosis, and acute coronary syndrome, shedding light
on the complex phenotypic spectrum of this disorder. This report seeks to elucidate the clinical course, diagnostic challenges, and therapeutic considerations in such a unique presentation.

METHODS:

CASE REPORT:

36/M, unmarried with no prior comorbidities or addictions presented with chief complaints of worsening bipedal edema for 6 months associated with abdominal distension and darkening of skin. Patient also gave history of intermittent melena and hemorrhoids. Examination revealed icterus, shifting dullness and skin hyperpigmentation .Blood investigations revealed thrombocytopenia, deranged liver function tests. Ultrasonography suggested chronic parenchymal liver disease with moderate ascites. Workup for the etiology of CLD revealed elevated iron indices, high SAAG low protein ascites and negative markers for Wilson's disease, viral and autoimmune hepatitis.On the 5th day of admission, patient developed typical cardiac chest pain with ST segment depression on leads I, aVL and elevated troponins.
Genomic analysis revealed heterozygous mutation in GBA gene c509G>A suggestive of Gaucher disease.The patient was managed conservatively with standard medications for decompensated CLD and acute coronary syndrome. Ascites responded to diuretics. Patient is currently enrolled for enzyme replacement therapy and is better now.

INVESTIGATIONS

Complete Blood - Count 6600/ 11.6/ 67000 per mm3
Random Blood Sugar - 94 mg/dl
Total / Direct Bilirubin - 2.9 / 1.2 mg/dl
AST/ALT - 86/53 mg/dl
Total Protein / Albumin - 5.5 / 2.5 mg/dl
ALP - 146 mg/dl
Creatinine - 0.8 mg/dl
HIV/ HBsAg/ Anti HCV ELISA : Non - reactive
Direct & Indirect Coomb's test - Negative
Peripheral smear - Normocytic normochromic anemia
Fasting cortisol - 14.87
Serum ferritin - 378 ng/ml
Fasting transferrin saturation - 45.5 %
ANA – IF - Negative
Autoimmune hepatitis antibody panel - Negative
Serum ceruloplasmin - 28.61 mg/dl
TSH - 0.95 mIU/ml
Serial EKG - ST depression in lead I, avL
qTroponinI -1.36 ng/ml
USG Abdomen Chronic parenchymal liver disease with
portal hypertension with splenomegaly and moderate ascites
Whole genome analysis - GBA (-), c509G>A, p.ARG170His, heterozygous mutation

MANAGEMENT :

Guideline directed management of Acute Coronary Syndrome and Decompensated Chronic Liver Disease.
Enrolled for enzyme replacement therapy.

CONCLUSIONS:

In conclusion, this case report underscores the heterogeneous nature of Gaucher disease and its potential to manifest with a wide spectrum of clinical presentations, including hepatic and cardiovascular complications.

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