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THE BIG SPLEEN MYSTERY: GAUCHER DISEASE AS A DIAGNOSTIC SURPRISE IN AN ADOLESCENT - A RARE CASE REPORT

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ABSTRACT Gaucher disease is the most common lysosomal storage disorder caused by deficiency of the enzyme glucocerebrosidase, resulting in accumulation of glucocerebroside within macrophages. It is inherited in an autosomal recessive pattern and commonly presents with hepatosplenomegaly, cytopenias, and skeletal involvement. We report a 16-year-old female who presented with abdominal distension, pain, easy fatigability, and jaundice for 6 months. Examination revealed severe pallor, icterus, and massive hepatosplenomegaly. Laboratory investigations showed severe pancytopenia with elevated bilirubin levels. The patient developed congestive cardiac failure secondary to severe anemia and was stabilized with blood transfusion. Bone marrow aspiration and biopsy demonstrated characteristic Gaucher cells, confirming the diagnosis. This case highlights the importance of considering storage disorders in adolescents presenting with unexplained pancytopenia and organomegaly. Early diagnosis is essential for timely initiation of enzyme replacement therapy and prevention of long-term complications.

KEYWORDS : Gaucher Disease, Pancytopenia, Hepatosplenomegaly, Lysosomal Storage Disorder, Adolescent

INTRODUCTION

Gaucher disease is the most common lysosomal storage disorder caused by deficiency of the enzyme glucocerebrosidase, leading to accumulation of glucocerebroside in macrophages. It is inherited in an autosomal recessive pattern and manifests with hepatosplenomegaly, cytopenias, and skeletal involvement.[1] Early diagnosis is crucial to prevent long-term complications and initiate enzyme replacement therapy. We report a rare presentation of Gaucher disease in an adolescent with pancytopenia and congestive cardiac failure.

Case Study

A 16-year-old female presented with complaints of abdominal pain, progressive abdominal distension, easy fatigability, and yellowish discoloration of eyes for the past 6 months. There was no history of fever, bleeding manifestations, or joint pain.

On examination, she had tachycardia, tachypnea, and raised jugular venous pressure. Severe pallor and icterus were evident. Abdominal examination revealed significant hepatosplenomegaly: liver palpable 7 cm below the right costal margin (span 14.5 cm), and spleen palpable across the midline into the right iliac fossa (span 17 cm), both of firm consistency.

Laboratory evaluation showed:
Severe pancytopenia: Hb 3.6 g/dL, WBC 1410/cu.mm, ANC 750/cu.mm, Platelets 76,000/cu.mm
Liver function tests: Elevated total bilirubin 2.0 mg/dL
Peripheral smear: Microcytic hypochromic anemia with anisopoikilocytosis, no atypical cells

In view of anemia with signs of congestive cardiac failure, she was stabilized with a packed red blood cell transfusion (5 ml/kg over 6 hours). A provisional diagnosis of pancytopenia with massive splenohepatomegaly was considered, with differential diagnoses including hemato-lymphoid malignancy and storage disorders.

Bone marrow aspiration and biopsy were performed, which revealed infiltration by lipid-laden macrophages ("Gaucher cells"). Based on morphological findings, a diagnosis of Gaucher disease was made. Enzyme assay and genetic testing are awaited to confirm the diagnosis.

DISCUSSION

Gaucher disease is caused by mutations in the GBA1 gene, leading to deficient lysosomal glucocerebrosidase activity. The accumulation of glucocerebroside in macrophages results in characteristic Gaucher cells infiltrating the bone marrow, liver, and spleen. Type 1 (non-neuronopathic) Gaucher disease is the most common form and can present with anemia, thrombocytopenia, and massive organomegaly.[1,2]

This case is notable for its presentation in late adolescence, which is relatively rare, and for the severity of symptoms including congestive cardiac failure due to profound anemia. The diagnosis is often delayed due to its nonspecific presentation and overlap with other hematological disorders.[3]

Diagnosis is confirmed with enzyme assay (reduced glucocerebrosidase activity) and genetic testing. Treatment includes enzyme replacement therapy (ERT), which significantly improves hematological parameters and organomegaly.[4,5]

Early recognition and initiation of treatment are essential to improve long-term outcomes and prevent irreversible complications such as bone crisis or pulmonary involvement.

CONCLUSIONS

Gaucher disease, though rare, should be considered in adolescents presenting with unexplained pancytopenia and massive hepatosplenomegaly. Bone marrow biopsy can offer important clues, but confirmation requires enzymatic and molecular tests.

Early diagnosis and appropriate management with enzyme replacement can significantly improve quality of life and prognosis

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