Gaucher's disease in a child with Isolated Splenomegaly - An unusual clinical presentation

Ishita Gupta*, Garima Sharma, Natwar Parwal, R.M. Jaiswal and Anjan a Mittal

Mahatma Gandhi University of Medical Sciences and Technology, Sitapura, Jaipur, Rajasthan 303905 India

*Correspondence Info:
Dr. Ishita Gupta
Mahatma Gandhi University of Medical Sciences and Technology,
Sitapura, Jaipur, Rajasthan 303905 India

*Article History:
Received: 26/01/2017
Revised: 31/02/2017
Accepted: 02/02/2017
DOI: https://dx.doi.org/10.7439/ijbar.v8i1.3906

Abstract
Gaucher’s disease is an autosomal recessive, lipid storage (sphingolipid) disorder caused by mutations in the beta-glucocerebrosidase gene leading to deficient activity of enzyme. There are three types of Gaucher’s disease out of which Type 1 Gaucher’s disease is most common and usually presents as Splenomegaly, hepatomegaly and radiological bone disease. Here we are reporting a case of type-I Gaucher’s disease presenting with isolated splenomegaly at a younger age (2 ½ year).

Keywords: Gaucher’s disease, sphingolipid, Splenomegaly.

1. Introduction
Gaucher's disease is an autosomal recessive, lipid storage (sphingolipid) disorder caused by mutations in the beta-glucocerebrosidase gene leading to deficient activity of enzyme. There are three types of Gaucher's disease out of which Type I Gaucher's disease is most common and usually presents as Splenomegaly, hepatomegaly and radiological bone disease. Here we are reporting a rare case of type-I Gaucher's disease presenting with isolated splenomegaly at a young age (2½ year).

2. Case Report
A 2½ year old female child came to us with history of abdominal fullness and growth retardation since 6 months of age. She never had any history of bleeding tendencies, bone and joint involvement or recurrent fractures. There was family history of one sibling death at the age of 2.5 years in past. On abdominal examination spleen was palpable, 3 cm below the costal margin, however liver was non palpable.

Abdominal ultrasound showed massive splenomegaly, measuring 15.2 cm in size. Other systemic examinations were within normal limits. Fundus examination showed a characteristic cherry red spot. Her CBC and Peripheral smear examination showed microcytic hypochromic anemia with thrombocytopenia. Other investigations viz LFT, RFT and Hb electrophoresis were normal. In view of suspicion of a storage disorder her Bone Marrow examination was done which showed mild erythroid hyperplasia with features of dyserythropoiesis and dysmegakaryopoiesis.

Few large histiocytic cells resembling gaucher cells were seen having crumpled tissue paper appearing cytoplasm. These cells were PAS positive and therefore gave impression of Gaucher's disease. Liver biopsy also showed gaucher cells consistent with Gaucher's disease. Enzyme analysis showed deficiency of beta-glucosidase, an enzyme consistent with the diagnosis of Gaucher's disease.
3. Discussion

Gaucher disease (G.D.) is a lipid storage disease characterized by the deposition of glucocerebroside in cells of the macrophage-monocyte system. It was first described by Gaucher in 1882, and the storage of glucocerebroside was first recognized by Epstein in 1924. The metabolic defect, which is the deficiency of the lysosomal hydrolase B-glucosidase, or B-glucocerebrosidase, was identified by Brady et al.[1] Overall incidence is approximately 1:40,000.[2] The most common signs and symptoms noted in Gaucher’s disease are hepatosplenomegaly (95%), radiological bone disease (81%), thrombocytopenia (50%), anaemia (40%), growth retardation (34%), bone pain (27%) and bone crisis (9%).[3]

Enlargement of the spleen appears to be most rapid in children with Gaucher disease and splenomegaly is reported as most consistent finding with gaucher disease. Hepatomegaly occurs in more than 50% of patients with type I GD.[4,5] Though in a study performed by Laila Essabar in eleven cases of gaucher’s disease hepatosplenomegaly was observed in all the cases. Our case has isolated splenomegaly with no finding in hepatobiliary system.[6]
Bone disease has a very high prevalence in type 1 GD, with radiological evidence described in 93% of patients. Data from the International Collaborative Gaucher Group (ICGG) Registry shows that at diagnosis bone pain is present in 50% of type 1 GD patients. In our case there was no complaint of bone pain and all skiagrams of bones and joints were normal.

Hematologic manifestations of GD include pancytopenia with anemia, thrombocytopenia and less commonly leucopenia. Our case showed microcytic hypochromic anemia and thrombocytopenia with normal total leucocyte count. Suspicion raised towards metabolic storage disorder was suspected owing to the past family history.

The pathologic hallmark of GD is the presence of Gaucher cells in the macrophage-monocyte system, particularly in the bone marrow or in liver biopsy samples. Diagnosis can be confirmed through measurement of glucocerebrosidase activity in peripheral blood leukocytes. Bone marrow examination was done which showed Gaucher cells which were PAS positive. Liver biopsy was also done which showed Gaucher cells consistent with Gaucher's disease. Enzyme test for Gaucher's disease showed deficient activity of beta Glucosidase consistent with the diagnosis.

4. Conclusion

B.M. Examination, Liver Biopsy and Enzyme analysis are the hallmark for diagnosis of Gaucher's disease. Storage disorders specially Gaucher's should be kept in the differential diagnosis of any child presenting with isolated splenomegaly and growth retardation after ruling out other etiologies like Chronic Infection, EHPVO, Chronic liver disease, Hemolytic anemia, Malignancies etc. Early recognition of Gaucher's disease would lead to safe and effective treatment with enzyme replacement which can decrease morbidity and reduce visceral and skeletal involvement.

References