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An unusual case of Pancytopenia with Hepatosplenomegaly

Dr Farid Alam

Banaras Hindu University, India

Gaucher's disease is one of the most common lysosomal storage diseases characterized by hematologic abnormalities, organomegaly, and skeletal involvement. It is caused by reduced activity of the enzyme acid β-glucosidase which is encoded by a gene on chromosome 1q21-q311. The enzymatic defect results in the accumulation of glycolipid substrates, mainly glucosylceramide, in cells of the macrophage-monocyte system. It is one of the most prevalent genetic defects among Ashkenazi Jews2. There are 3 clinical subtypes distinguished by the presence or absence and progression of neurologic manifestations: type 1 or the adult, non neuronopathic form; type 2, the infantile or acute neuropathic form; and type 3, the juvenile or sub-acute neuropathic form3.

Here we report a case of Gaucher's disease that presented with pancytopenia and splenomegaly. We present this case to emphasize the importance of considering storage disorders like Gaucher's disease when evaluating a case of unexplained pancytopenia and organomegaly even in adults.

DISCUSSION

Gaucher's disease is an autosomal recessive disorder that affects all racial and ethnic groups with maximum prevalence among Ashkenazi Jews. It is characterized by the deficiency of the enzyme glucocerebrosidase which results in accumulation of glucocerebroside in the cells of reticuloendothelial system. It is classified into three clinical subtypes of which Type 1 is the non neuronopathic form and is the most common subtype. Hematological disorders were ruled out because of absence of important signs like jaundice, leukocytosis, lymphadenopathy and abnormal hemoglobin electrophoresis. Certain infectious disorders like HIV, kala azar, tuberculosis and malaria can present with massive splenomegaly but our patient did not have fever throughout the course of illness and these were subsequently ruled out on the basis of serological tests. Infiltrative disorders like sarcoidosis and certain storage disorders can present with organomegaly and pancytopenia. Sarcoidosis was ruled out by normal serum angiotensin converting enzyme level and absence of lung disease. The clinical symptom profile of our case was consistent with Gaucher's disease and bone marrow examination showing gaucher cells supported our diagnosis. All suspected cases, however, must be confirmed by demonstrating decreased acid β-glucosidase activity in isolated leukocytes as pseudo-Gaucher cells can be seen in many hematological malignancies and AIDS.

Biography

Dr Farid Alam is a General Medicine professor at Banaras Hindu University, India

faridalam011gmail.com

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