An Unusual Case of Adult Type Gaucher’s Disease

Maria Shafiq*, Ayesha Danish*, Hamid Iqbal**
*Department of Pathology Army Medical College Rawalpindi; **Department of Pathology Military Hospital Rawalpindi

Introduction

Gaucher’s disease is lysosomal storage disorder, autosomal recessive in inheritance. Due to the mutation of glucocerebrosidase gene, there is deficiency of the enzyme glucocerebrosidase responsible for the degradation of sphingolipid, glucocerebrosid. Different clinical manifestations appear as a result of accumulation of glucocerebrosid laden macrophages in various organs according to the type of disease. It is divided into three types. Type 1 is common in Ashkenzai jews. We report an unusual case of an adult female with type-1 Gaucher’s disease.

Case Report

A 27-year old female resident of Gojra presented with gestational amenorrhea of 8 weeks, abdominal distension and associated pain in left hypochondrium. On examination she was pale. Abdominal examination revealed tender and massive splenomegaly along with enlarged liver. Abdominal Ultrasonography confirmed hepatosplenomegaly without the evidence of portal vein thrombosis. Eye examination revealed bilateral pterygium. Other systems examination did not show any significant finding.

Her complete blood counts showed Hb 9.8 g/dl, white cell count 5.4 X 10^9/l (lymphocyte 26 %, neutrophil 70 %, eosinophils 2 %, monocytes 2 % and basophils 1% ) and platelets 70 X 10^9/l. Reticulocyte count was 4%. Hypochromic microcytic picture with numerous pencil cells were seen on peripheral blood film. Liver and renal function test were within normal limits. Hepatitis serology was negative. LDH was not raised.

One unit red cell concentrate was transfused and bone marrow was advised for evaluation of hepatosplenomegaly. Bone marrow aspiration showed hypercellular marrow with megaloblastic changes and typical Gaucher cells. Findings of trephine biopsy were consistent with that of aspirate and confirmed the diagnosis of Gaucher’s disease. She was given folic acid and was discharged.

Discussion

Type 1 Gaucher’s disease is the most common type and has been named adult type. But 60% of individuals show onset of symptoms in the first two decades of life. Incidence is 1 in 45,000-60,000 in the general population and 1 in 850 among Ashkenazi Jews.

This type of Gaucher’s disease has varied symptoms and signs depending on the organ involved at the time of presentation. Hepatosplenomegaly, pancytopenia due to bone marrow suppression and bone lesions are the common features. Pulmonary and renal involvement can also be there.

The disease is caused by mutation in lysosomal glucocerebrosidase gene on the first chromosome (1q21). Inspite of variance in age and mode of presentation all patients carry the p.N370S mutation.

Although the mean age of presentation of this
disease is 27.2 years according to an international study but in Pakistan, case reports reveal that generally these patients present before the age of two years. A study done recently in our setup also support this finding.

In this case patient presented at the age of 27 years supporting the mean age of presentation observed in studies done at international level. It raises the importance of investigating the causes of splenomegaly at all ages. As enzyme assays are neither available nor affordable for the common people of Pakistan, the reliance is totally upon bone marrow examination for diagnosis. Cytochemical stains PAS and ACP are used to confirm the presence of Gaucher's cell and to differentiate it from other storage cells.

A recombinant DNA modified form of glucocerebrosidase, known as Imiglucerase is the choice of treatment in such patients. It is given at regular intervals at a dose of 60μg/kg, once every 2 weeks. It decreases splenic size, reduces the requirements for transfusions and improves any cardiopulmonary symptoms present. This enzyme replacement therapy is not yet available in Pakistan. However, it is provided to the patients on demand, hence is a burden for the resources of the country and very costly for the patients. Miglustat, an oral drug can provide benefit to patients who do not respond to enzyme replacement therapy. Bone marrow transplant is the only curative option that can increase the life expectancy.

References