Research Article

Gaucher's Disease: A Case Report

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ABSTRACT

Gaucher disease is an inherited disorder that affects many of the body's organ and tissues. It results from not having enough glucocerebrosidase, important enzyme that breaks down fatty chemical called glucocerebroside. This disease is a multisystem lipidosis characterized by hematological changes, organomegaly and skeletal involvement. Blood test should be done to measure the amount of enzyme glucocerebrosidase and compares it to normal enzyme activity levels. Counseling is encouraged to help families manage some of emotional stability.

Keywords: Gaucher, disease, enzyme.

INTRODUCTION

Gaucher disease is a rare genetic disorder results from the deficiency of the enzyme glucocerebrosidase also termed (acid beta glycosidase, glucosylceramidase).[1] It is pronounced "go shays".[2] It was first described by Gaucher in 1882, and the storage of Glucocerebroside was first recognized by Epstein in 1924.[3] Gaucher's disease isn't something you catch like cold or the flu. The main cause of the Cytopenia, Spleenomegaly, hepatomegaly and bone lesions associated with the disease is considered to be infiltration of the bone marrow, spleen and liver by Gaucher cells. Symptoms of type 1 can appear at any time in your life as bruising, nosebleeds, fatigue and enlarged spleen or liver, bone problems like arthritis. Type 2 affects both brain and spinal cord and is very serious as slow back and forth eye movement, failure to thrive. High pitched sound when breathing, seizures, and brain damage. Type 3 also affects both brain and spinal cord symptoms are similar to type 2. Enzyme replacement therapy is one option for type 1 and 3. It also reduce anemia and shrinks an enlarged spleen or liver. Medications like Imiglucerase (Cerezyme), Taliglucerase alfa (Elelyso), Velaglucerase alfa (VPRIV). There's no treatment that can stop type 3 from causing damage to the brain. Gauchers is a progressive disease which means it develops gradually. [4]

Case Report

When I took B.Sc nursing III year students for the Paediatric Nursing Clinical experience in SRM General hospital, kattankulathur, I have got a chance to observe and care a 2 years old toddler with Gaucher's disease. The parents brought the child to the paediatric medical ward with the following complaints Abdominal distension (abdominal girth =53cm) with significant hepatomegaly (since 6 months), splenomegaly, weight loss (10 kg), pallor, yellowish discoloration of the skin and conjunctiva. On admission, heart rate was 48 breaths/min, and BP was 100/50 mmHg. The mother expressed that her child was not taking food properly since 2 weeks. parents had consulted many paediatricians in their home town , but no cause had been found The parents were very anxious because their experience with health care settings is limited to well child checkups which were scheduled a two weeks before. After a thorough history is taken, blood tests are performed to assess the cause. haemoglobin value is 7.7 gm (anemia) , total WBC count is 7,100 cell/cumm, Neutrophils 33 %, lymphocytes 60 %, eosinophills 04 %, monocytes 03%, Total RBC count 3.7, total platelet count is 89,000 (Thrombocytopenia) , MCV 70, MCH 21, MCHC 29.

DISCUSSION

The result of Ultrasonography is spleen (enlarged in size 15.4 cm), Gall bladder (contracted), and liver (hepatomegaly). And peripheral hypochromic anemia with thrombocytopenia. On the basis of abdominal ultrasonography and blood tests, the child was diagnosed with Gaucher's disease. The child was admitted to the paediatric intensive care unit so that the child can be monitored and prevented from the threatening. A multi disciplinary team, including an intensive nurse, paediatric hepatologist, general physician, and anesthetist and social service professionals were involved in planning the care. The child had Antibiotic prophylaxis (Augmentin 625 mg) and vitamin supplements (Zincovit).I was helping the parent to apprehend the severity of the disease and to take up the treatment responsibility. I and my students were provided the following care under the guidance of PICU staffs. We had explained about the importance of initial tests during diagnostic evaluation. Differential diagnosis of Gaucher disease must be kept while dealing with child having massive Hepatospleenomegaly.[5]

CONCLUSION

Health education was delivered to the parents regarding the child's condition, care of the skin, timely administration of medications, measuring abdominal girth timely, monitoring for increasing symptoms of liver disease, meeting nutritional needs based on the child's age including special formulas , vitamins and mineral supplements and need for follow up . Moreover the early recognition of Gaucher disease would lead to safe and effective treatment with enzyme replacement which can decrease morbidity and reduce as far as possible the visceral and skeletal involvement.[6]

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