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Research Article

PYREXIA OF UNKNOWN ORIGIN -AN ATYPICAL PRESENTATION OF INFANTILE GAUCHER'S DISEASE

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ABSTRACT

Gaucher's disease is an autosomal recessive lipid storage disease characterised by deposition of glucocerebroside in cells of macrophage – monocyte system. The metabolic defect is due to the deficiency of the lysosomal hydrolases β-glucosidases. Incidence is approximately 1 in 40,000. An one year female child presented with history of prolonged fever, pallor and huge hepatosplenomegaly for 25 days. Clinical diagnosis of leukaemia was made. Routine haemogram reveals microcytic hypochromic anaemia. Ultrasound of abdomen and pelvis reveals hepatomegaly with fatty change and splenomegaly. Endoscopy reveals dilated veins at lower end of esophagus (? Early varix). Bone marrow aspiration study reveals normal haematopoesis admixed with distended macrophages in good number with wrinkled tissue paper like cytoplasm (? Gaucher's cell). β glucocerebrosidase level found to be 0.9 unit (low) confirming the diagnosis of Gaucher disease.

INTRODUCTION

Gaucher's disease is an autosomal recessive lipid storage disease, characterized by accumulation of glucocerebroside in organ and tissue as a result deficiency of lysosomal glucocerebrosidase. It is characterized by presence of gaucher's cells in bone marrow, liver, spleen and lymph node. Its incidence is approximately 1 in 40,000. It was first described by Gaucher in 1882, and therefore the storage of glucocerebroside was first recognized by Epstein in 1924. The metabolic defect, which is that the deficiency of the lysosomal hydrolase ß-glucosidase, or ß-glucocerebrosoidse, was identified by Brady et al .[1] There are threesubtypes, basing on the absence or presence of neurologic involvement : type 1 or the non-neuropathic form, type 2, the infantile-onset, acute neuronopathic form and type 3, the juvenile- onset neuronopathic form [2]. The pattern of inheritance of three subtypes are autosomal recessive mode. Type 1 disease is that the commonest lysosomal storage disease and one among the foremost prevalent genetic disorders among Ashkenazi Jewish individuals with an incidence of about 1 in 450. [3]

Here we report a case of Gaucher disease presented with prolonged fever, pallor and massive splenomegaly as the prodromal symptoms. We presented this case to emphasize the importance of clinical examination and bone marrow finding in the diagnosis of Gaucher disease.

Case report

One year female child presented with history of fever since 25days. Parents were also complaining of gradually increasing abdominal girth. She was a Hindu girl and born with a parents with no consanguineous marriage. She was delivered after full-term normal pregnancy. Development of the child was normal. The patient's medical history was not significant.

On admission, the patient looked thin, ill and febrile. On physical examination, moderate degree of pallor was noted; however there was no icterus or lymphadenopathy. He had massive nontender splenomegaly and a mild nontender hepatomegaly. There were no signs of neurological abnormalities. Rest of systemic examination was essentially normal. Lab investigations revealed anaemia with haemoglobin was 8.2 g/dl (Fig 3) while total leucocyte count & platelet count were within normal limit. Liver enzymes were increased slightly to moderately but serum proteins and albumin, kidney function test, coagulogram and urine analysis were unremarkable. Hemoglobin electrophoresis was found to be normal. All investigation for fever was performed and came

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negative. Ultrasound revealed grossly enlarged spleen (118 x 47mm) (Fig 1). Liver span was 99mm with mild fatty change (Fig 2). Endoscopy report reveals dilated veins at lower end of esophagus (Early varix).

To evaluate prolonged fever with massive splenomegaly, bone marrow aspiration was performed which revealed normal hemotopoietic cells with additional findings of distended macrophages in good number with wrinkled tissue paper like cytoplasm (Gaucher's cells) (Figure 4,5**).** Confirmation of diagnosis on Gaucher's disease (type 1) was performed by unit of beta glucosidase levels - 0.9 nmol/hv/mg (normal levels >6.0). Final diagnosis of Gaucher disease (type-1) was made.

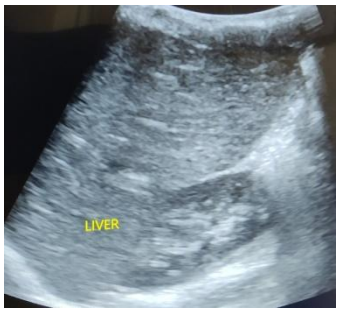

Fig. 1 Photomicrograph: Ultrasonography showing hepatomegaly

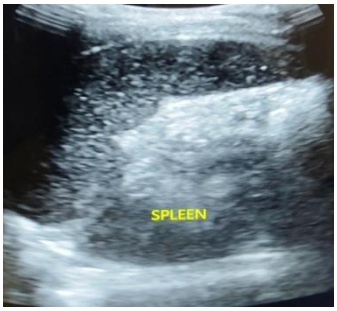

Fig. 1 Photomicrograph: Ultrasonography showing huge splenomegaly

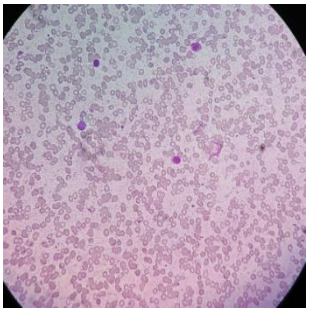

Fig. 3 Photomicrograph: Peripheral smear (100x) showing anaemia

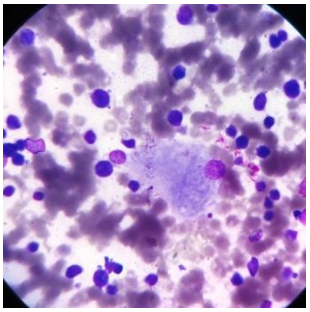

Fig. 4 Photomicrograph: Bone marrow (400x) showing distended macrophages with wrinkled tissue paper like cytoplasm (Gaucher's cells)

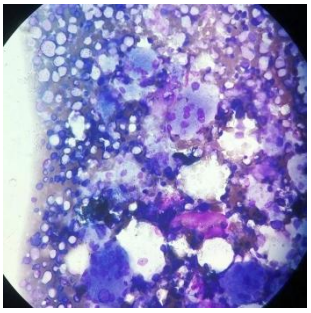

Fig. 5 Photomicrograph: Bone marrow (400x) showing good number of Gaucher's cells

DISCUSSION

Gaucher's disease is an autosomal recessive lipid storage lysosomal disorder.[4] Its prevalence is higher in Ashkenazi Jews, however can be seen in any population. It affects multiple organ systems. The amount of lipid stored, nor the residual enzymatic activity detected, correlates well with symptoms severity. [6]About two-thirds of the GD patients present before the age of 20 and onset in childhood is predictive of severe and progressive phenotype. [7]

Depending on the presence or absence of neurological involvement and its severity, GD has traditionally been classified into three types.[8] : Type 1 non-neuropathic form (chronic or adult form), Type 2: neuropathic form (acute or infantile form) and Type 3 : neuropathic form (subacute or juvenile form). The main clinical manifestations of type 1 GD includes splenic and skeletal involvement, that is why diagnosis is delayed. [1] The cytopenias are secondary to hypersplenism and infiltration of the bone marrow by Gaucher cells.

In most patients, GD is diagnosed by hematologists, usually by means of a bone marrow trephine biopsy.[1] Gaucher cells are usually large and can measure up to 100 microns in diameter; they may have one or more dark eccentric nuclei and rarely have vacuoles in their cytoplasm. With the electron microscope it is understood that the striated aspect of the cytoplasm is due to elongated lysosomes loaded with lipids.[4] The differential diagnosis of "foamy" macrophages in bone marrow should are: Gaucher disease, Fabry disease, Gangliosidosis GM1, Niemanne Pick A and B and hematologic disorders with pseudo-Gaucher cells.

When GD is suspected, diagnosis is confirmed by measuring the beta-glucocerebrosidase activity in the blood leukocytes. The diagnosis can be confirmed by a molecular study.[1] More than 300 different mutations for GD have been documented by the Human Gene Mutation Database.

Enzyme-directed macrophage replacement therapy (TRE) has long been the quality treatment. It doesn't repair the underlying genetic disease but it can reverse and stop numerous manifestations of type 1 GD. Our patient, at the moment of this publication has so far been managed following these general measures like blood transfusion. However, patient expired due lack of specific treatment. The present case is of relevant interest to the scientific community thanks to its low incidence, which makes it a rarely-suspected diagnostic possibility, leading, not infrequently, to a diagnostic delay, which makes it important for medical personnel responsible of its diagnosis and monitoring.

CONCLUSION

G.D. should be considered within the medical diagnosis of patients with unexplained splenomegaly especially with an extended period of your time . Moreover, the first recognition of GD would cause safe and effective treatment with enzyme replacement which may decrease morbidity and reduce as far as possible the visceral and skeletal involvement.

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