Case Report

DOI: http://dx.doi.org/10.18203/2349-3291.ijcp20182588

Gaucher disease: masquerading as chronic malaria

Seema Rai*, Gurmeet Kaur Sethi, Rama Kumari, Varun Kaul

Department of Paediatrics, Guru Gobind Singh Medical College, Faridkot, Punjab, India

Received: 19 March 2018 Accepted: 27 April 2018

*Correspondence:

Dr. Seema Rai,

E-mail: seemadoc98@yahoo.co.uk

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ABSTRACT

Gaucher disorder s rare lysosomal disorder characterized by glycolipid laden lysosomes leading to hepatosplenomegaly, bone marrow involvement. Three types of Gaucher disease have been described based on the clinical features, ethnicity and the natural history of the disease. Gaucher disease Type 1 (GD1) occurs mainly in infancy to adulthood and is the commonest lysosomal storage disorder. Gaucher Disease Type II (GD2) and Gaucher disease type III (GD3) patients have onset at less than 1 year, and 2-20 years, respectively.1 GD1 patients do not have neurological involvement. GD2 is the acute neuronopathic and GD3 is the chronic neuronopathic type.

Keywords: Gaucher, Glycolipid, Hepatosplenomegaly, Lysosomal

INTRODUCTION

Gaucher disease is a rare lysosomal disorder characterized by haematological abnormality, organomegaly and skeletal involvement, served as a prototype for genetic/phenotypic delineation as well as therapeutic innovation.

The overall incidence is about $1:40000.^1$ The metabolic defect is an inherited deficiency of lysosomal acid β -glucosidase due to mutations in the GBA1 gene. This results in accumulation of glucocerebroside in lysosomes of macrophages involving liver, spleen, bone marrow and lungs.²

Gaucher disease is classified into three broad phenotypic categories depending on the absence or presence of neurological involvement; although overall it represents a continuum of disease spectrum.^{3,4}

An increased incidence of neuronopathic Gaucher disease has been reported from other countries but the frequency and phenotypic spectrum of Gaucher disease in India is currently unknown.^{5,6} There have been isolated case reports of GD in India.^{7,8}

CASE REPORT

A 2 year old male presented with abdominal distension for 11 months, gradual in onset, progressive in nature associated with weight loss, born to parents with nonconsanguineous marriage, cried immediately after birth, mental-motor milestone were normal for age, family history non-contributory and immunized per age, child was treated at periphery for malaria received two courses of antimalarial treatment. Physical examination revealed underweight, pallor and bilateral anterior and posterior non-tender cervical lymphadenopathy, splenohepatomegaly.

Laboratory investigations revealed haemoglobin 4.5 g/dl, white count blood cell count 4.6×103/uL with 60% lymphocytes, 38% neutrophils and platelets 50000/cumm. Ultrasound examination showed hepatomegaly with altered echo texture, splenomegaly with altered echo texture, bone marrow examination

showed gaucher cells (macrophage with glycolipid) as shown in Figure 1 and 2.

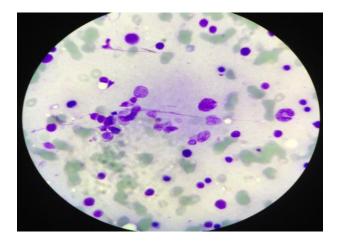


Figure 1: Enlarged macrophage.

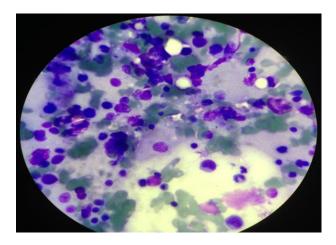


Figure 2: Increased cellularity in background and macrophages showing crushed paper appearance.

DISCUSSION

Gaucher disease was first described by Philippe Gaucher in 1882. Gaucher diseae is an autosomal recessive disorder of metabolism where glucocerebroside cannot be adequately degraded. Gaucher disease is characterized by splenomegaly (95%), hepatomegaly (87%), radiological bone disease (81 %), thrombocytopenia (50%), anemia (40%), growth retardation (34%), bone pain (27%), and bone crisis (9%).

Three types of Gaucher disease have been described based on the clinical features, ethnicity and the natural history of the disease. Gaucher disease Type 1 (GD1) occurs mainly in infancy to adulthood and is the commonest lysosomal storage disorder. Gaucher Disease Type II (GD2) and Gaucher disease type III (GD3) patients have onset at less than 1 year, and 2-20 years, respectively. GD1 patients do not have neurological involvement. GD2 is the acute neuronopathic and GD3 is

the chronic neuronopathic type. On microscopy, Gaucher cells are positive with PAS, Oil red O.

This highly aggressive phenotype with spleno-hepatomegaly, cytopenia, irritability, bone involvement and failure to thrive is associated with early mortality without treatment. The common differential diagnosis of the most prevalent presenting phenotype of splenohepatomegaly in Gaucher disease include hemolytic anemias typically hemoglobinopathies, non-cirrhotic portal hypertension, tropical splenomegaly, lymphoreticular malignancies and other storage disorder.

Funding: No funding sources Conflict of interest: None declared Ethical approval: Not required

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Cite this article as: Rai S, Sethi GK, Kaul V. Gaucher disease: masquerading as chronic malaria. Int J Contemp Pediatr 2018;5:1688-9.