International Journal of Medical and Pharmaceutical Research

Online ISSN-2958-3683 | Print ISSN-2958-3675
Frequency: Bi-Monthly
Available online on: https://ijmpr.in/

Case Study

Hemolytic Anemia Mimicker Came Out to be Gaucher Disease

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OPEN ACCESS

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Received: 09-12-2025
Accepted: 26-12-2025
Available online: 11-01-2026

ABSTRACT

Gaucher Disease is a metabolic disorder, inheritance being autosomal recessive. It is the most common amongst Lysosomal storage diseases. It is caused by deficiency of acid Glucocerebrosidase(GBA) enzyme. Consequently accumulation of cerebroside occurs in phagocyte cells in reticuloendothelial system and in some variants of the disease in CNS. There are three subtypes, most common(99%) being type 1(non-neuronopathic form) presenting as hepatosplenomegaly, pancytopenia, failure to thrive and skeletal complications. Type 2,3 manifesting with neurologic symptoms. Diagnosis is by bone marrow aspiration or biopsy, enzymatic assay of beta-glucosidase and confirmation by genetic testing.

We describe a 3year3 month old male child who presented with gradual abdominal distension over a period of 18 months. Examination revealing hepatosplenomegaly with pallor and jaundice, failure to thrive. Haemolytic anemia was first thought of till HPLC came to be normal. Differentials were narrowed down to infiltrative disorders likely storage disorders like Gaucher disease, Niemann Pick disease. Possibility of leukemia, HLH was remote due to absence of fever, lymphadenopathy. Enzymatic assay depicted a normal sphingomyelinase level and lower normal beta glucocerebrosidase level, thus ruling in Gaucher disease. Bone marrow aspiration study showed Gaucher cells. Child presently receiving conservative management with initiation of process to receive enzyme replacement therapy(ERT).

Thus Gaucher disease should be considered a differential , not uncommon in patients with unexplained organomegaly, jaundice with anemia or pancytopenia.

Keywords*: Hepatosplenomegaly, anemia, pancytopenia, Gaucher Disease, Gaucher cells, GBA enzyme, enzyme replacement therapy.*

INTRODUCTION

Gaucher disease is a multisystemic lipidosis, resulting from deficient activity of lysosomal hydrolase acid beta glucosidase, encoded by gene GBA. The enzymatic defect causes accumulation of undegraded glycolipid substrates in reticuloendothelial system with progressive infiltration of bone marrow, hepatosplenomegaly and skeletal complications.

There are three clinical subtypes on basis of absence or presence of neurologic manifestations- type 1, most common form(adult/ non neuronopathic), type 2 is infantile or acute neuronopathic form and type 3 is juvenile or subacute neuronopathic form. All being autosomal recessive in inheritance.

Type 1 GD presents with bruising from thrombocytopenia, chronic fatigue secondary to anemia, hepatomegaly with or without elevated LFT, splenomegaly and bone pain. Amongst skeletal manifestations, pathological fracture is common. Others being compression fracture of vertebral bodies, Erlenmeyer flask appearance of femur. Pulmonary infiltration may result in pulmonary failure. It is a premalignant state predisposing to lymphoproliferative diseases. Gaucher cell being pathologic hallmark have a characteristic wrinkled paper appearance due to intracytoplasmic substrate inclusions.

Type 2 GD characterised by rapid neurodegenerative disease and type 3 being classified as type 3a and 3b based on extent of neurologic disease-dementia and progressive myotonia(type 3a), isolated supranuclear gaze palsy(type 3b).

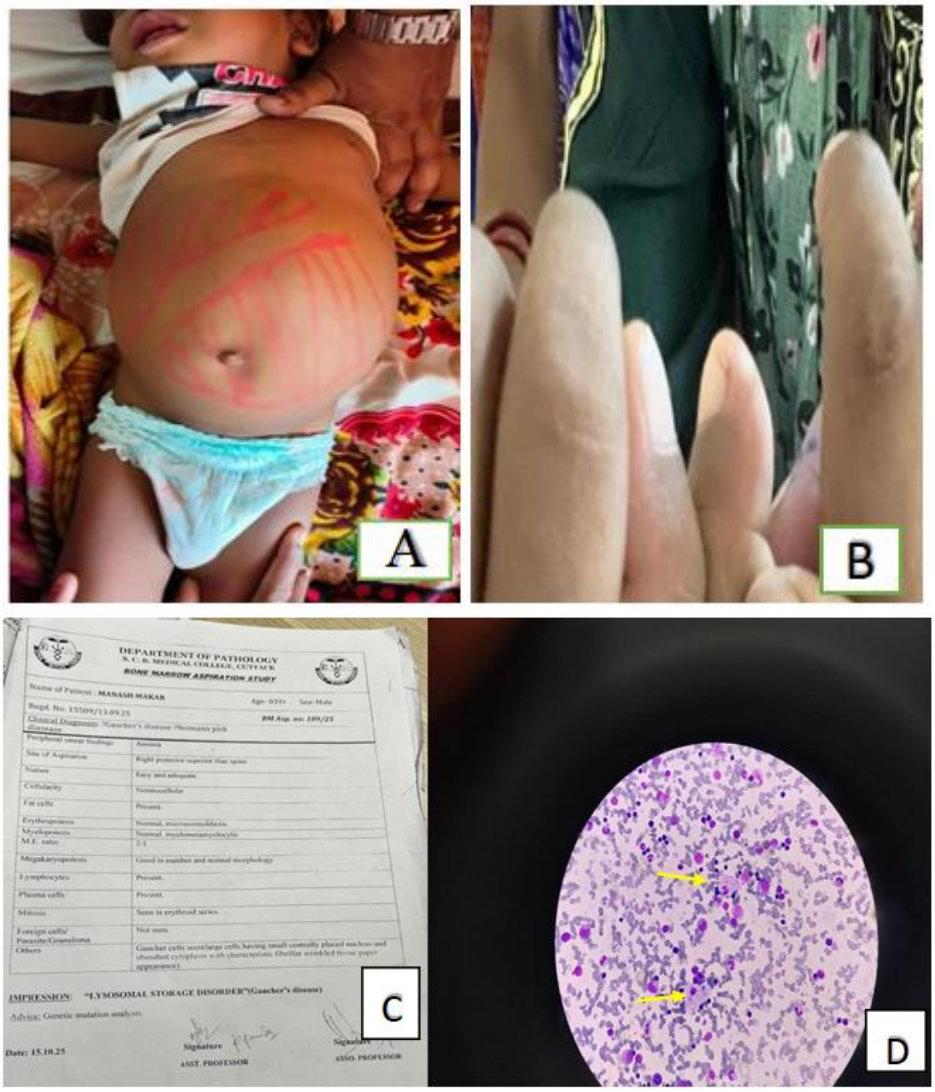
Diagnosis of type 1 GD can be done by bone marrow aspiration or biopsy showing gaucher cells, whereas definitive diagnosis of all types of GD requires assay of enzyme-acid beta glucosidase. Confirmation of diagnosis can be done by genetic testing.

Treatment of GD – Type 1 includes enzyme replacement therapy with recombinant acid beta glucosidase(Imiglucerase) which attenuates both skeletal as well as extraskeletal symptoms(organomegaly, hematologic indices)ERT is ineffective in Type 2 disease.

Other treatment options include substrate reduction therapy, chaperones, Piracetam, hip replacement(for avascular necrosis).

CASE STUDY

We present a case of 3 year 3 month old male child, fourth order in birth(birth weight -2.7kg), born out of consanguineous marriage, who came to visit for progressive painless abdominal distension for 18 months. Physical examination revealed- a coarse facies with pallor and icterus. Anthropometry recorded as weight 10.7 kg(-2 to-3 SD), height 67cm(<-3SD), weight for height(+2 to +3 SD). On abdominal examination, non tender hepatomegaly with liver span 13cm with firm consistency, smooth surface, sharp border. Spleen was enlarged and measured to be 14cm along splenic axis with palpable splenic notch. On laboratory examination, Hemoglobin was 7.3mg/dl, other blood cell indices remaining normal. HPLC was normal, blood glucose level normal, elevated PT, aPTT,INR levels, transaminitis with increased direct fraction of bilirubin. Skeletal survey was normal. Sphingomyelinase activity was normal while glucocerebrosidase level was at low-normal value. Bone marrow aspiration study showed Gaucher cells with normal hematopoetic activity. Genetic study could not be performed because of financial constraints. The child was supplemented with appropriate vitamins, iron folic acid and has been enrolled to avail enzyme replacement therapy.



Name	: Master MANAS MAKAR		
Lab No.	: 483248154	Age	: 3 Years
Ref By	: Self	Gender	: Male
Collected	: 20/9/2025 2:34:00PM	Reported	: 27/9/2025 5:35:25PM
A/c Status	: P	Report Status	: Final
Collected at	: Walk in LPL Cuttack 3rd floor, Unit no. 5, Jhanjiri Mangala, Purighat, OSL Tower 3, Traffic Tower, Link Road, Bdambadi Chowk, Near Badambadi Bus Stand Cuttack 753012.	Processed at	: LPL-NATIONAL REFERENCE LAB National Reference laboratory, Block E, Sector 18, Rohini, New Delhi -110085

Test Report

Test Name	Results	Units	Bio. Ref. Interval
NIEMANN PICK DISEASE, QUANTITATIVE, BLOOD (Spectophotometry)			
Patient Value	6.47	nmol/hr/mg	>3.00
Control Value	8	nmol/hr/mg	

Impression-: Normal Sphingomyelinase enzyme activity

Interpretation

SPHINGOMYELINASE ACTIVITY	REMARKS
>3	Normal activity
1.5-3	Possibility of carrier state likely
<1.5	Deficient activity

Note

1. Results should be clinically correlated as individual / biological variations can affect the test results
2. Genetic counseling available with prior appointment at Department of Genetics, National Reference Lab, New Delhi

Comments

Niemann Pick disease (Types A & B) is a lysosomal storage disease caused by deficiency of enzyme Sphingomyelinase. It is inherited as an autosomal recessive disorder.
Type A disease is characterized by jaundice, progressive loss of motor skills, feeding and learning difficulties and hepatosplenomegaly. It usually presents within 1-4 months of age and death occurs by 3 years.
Type B disease is milder though variable in clinical presentation. Most of these patients do not have neurological involvement and survive upto adulthood.
Type C disease is a lysosomal lipid storage disease presenting usually in middle to late childhood. It is characterized by vertical gaze palsy, ataxia, dystonia, behavioural problems and dementia.

E

GAUCHER DISEASE, QUANTITATIVE, BLOOD (Fluorometry)			
Patient Value	5.05	nmol/hr/mg	5.00 - 22.00
Control Value	6	nmol/hr/mg	

Fig A - hepato- & splenomegaly;

B- clubbing;

C- Bone marrow aspiration study suggestive of Gaucher's Disease;

D-Gaucher cells(yellow arrows) in bone aspiration slide examination;

E- lower normal beta glucocerebrosidase level.

DISCUSSION

In this case, normal HPLC; absence of fever, lymphadenopathy; absence of seizures and neuroregression; normal blood glucose level; normal skeletal survey ruled out possibilities of haemolytic anemia; hematologic malignancy; other storage disorders like Nieman Pick Disease, Glycogen storage diseases; Osteopetrosis.

Thus connecting consanguinity, coarse facies, age of presentation, chronic history, anemia, jaundice, hepatosplenomegaly in a 3 year 3 month old failure to thrive child with lower normal glucocerebrosidase level and Bone marrow aspiration study showing Gaucher cells points to diagnosis of Gaucher Disease Type1.
In case of anemia, jaundice, organomegaly the differentials to be thought of are-

1. Infections	Intrauterine infection in infants, Tropical splenomegaly, Disseminated Tuberculosis
2. Hemolysis and extramedullary hematopoesis	
3. Infiltrative	Non-neoplastic-storage diseases, osteopetrosis,HLH Neoplastic- Leukemia, LCH, Juvenile Myelomonocytic Leukemia
4. Liver Disease	
5. Immune Dysfunction	SLE, CVID,Sjia

Any under 5 child who presents with pallor, hepatosplenomegaly without fever after ruling out congenital haemolytic anemia, one has to bring the possibility of Gaucher Disease. Early diagnosis, timely ERT can lead to eventless outcome.

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