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CASE REPORT

Niemann-pick disease: Type A presenting as a case of protein energy malnutrition

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Author`s Contribution

[1] Manuscript presentation, follow up of patient
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A B S T R A C T

We hereby report a rare case of a young child with Niemann-Pick disease who had multiple hospital admissions due to repeated gastrointestinal and respiratory tract infections. The disease is overall quite rare in our population however, our case highlights the fact that in any young child with repeated infections of unknown etiology, this condition should be considered and evaluated.

Keywords: *Niemann-pick, protein-energy malnutrition*

Introduction

Niemann-pick is a rare genetic disease. It is autosomal recessive in inheritance with deficiency of a lysosomal enzyme called acid sphingomyelinase.[1] This results in lysosomal accumulation of sphingomyelin into various body organs. It is classified into four classical variants or types. Type A presents at an early age occurs because of missense mutation causing absolute deficiency of sphingomyelinase and can be life threatening in early period of infancy. Symptoms include feeding difficulties, failure to thrive and recurrent respiratory tract infections due to reduced lung capacities. Affected children have hypotonia and diminished tendon reflexes.[1] In type B, there is hepatic impairment, low platelets count, lungs involvement and visual changes. Common symptoms include recurrent respiratory tract infections, jaundice and some children may develop cirrhosis.

Type C Niemann-pick disease is characterized by visceral involvement including enlargement of liver, splenomegaly, neurological impairment, abnormal eye movements and psychiatric manifestations.[2] Neurological involvement is more likely in Type C as compared to Type B.[3] Type D is currently recognized as variant of Type C and occurs due to accumulation of cholesterol and sphingomyelin. It's more common in Nova Scotia hence also termed as "Nova Scotia" variant. Treatment of Niemann pick disease involves multidisciplinary approach including treatment of complications and rehabilitation, till now no treatment has proven to be curative.[4]

Case report

A one and half years old male, diagnosed case of Niemann-pick disease type A presented to the OPD of tertiary care hospital in Islamabad with presenting complaints of vomiting and diarrhea for 1 day as shown in Figure 1. There were 2 episodes of vomiting with a small quantity of yellow vomitus, and contained milk with food particles. No fever and no abdominal pain was recorded. Diarrhea was characterized as watery, loose with a frequency of 8 to 10 episodes per day. There was no mucous or blood in stool. Past history revealed repeated hospital admissions due to respiratory tract infections and gastroenteritis.

The developmental milestones were delayed including delay in raising the head while in lying position, poor response while calling his name at age of one year, inability to get up from lying position without support. He was diagnosed as Niemann pick disease type A through genetic testing with a positive homozygous pathological variant identified in SMPD 1 gene in a blood sample. The genetic testing revealed that the concentration of the lyso-SM-509 biomarker was pathologically increased and activity of acidic sphingomyelinase was pathologically decreased.[5]

On General physical examination, patient was afebrile and lethargic. Blood pressure was 90/50 mmHg and pulse rate was 128/minute. He was dehydrated with presence of pedal edema. On examination of the abdomen, there was generalized abdominal distension. The spleen was palpable crossing the umbilicus. Bowel sounds were audible. The patient was then admitted to the hospital for further management and discharged after stabilization and correction of dehydration.

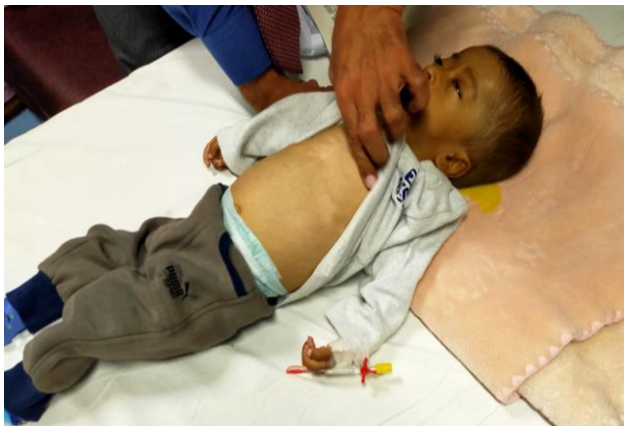

Figure 1: A one and half years old male, diagnosed case of Niemann-pick disease with the following investigation mentioned in Table 1.

Table 1: Investigation of patient

Investigations	Results
Hb	7.1
TLC	7900
Platelets	66000
CRP	16
Serum Albumin	<2.5
Serum Electrolytes: Na K Cl HCO3 BUN	 138 2.5 107 21 6
Cr	0.2
LFTs: ALT AST ALP	 367 60 105
Apt	49 Sec
PT	17.2

Discussion

The patient presented in our case report had multiple indoor admissions with repeated respiratory tract and gastrointestinal infections. His presenting clinical features especially splenomegaly, thrombocytopenia, anemia; delayed developmental milestones were suggestive of malnutrition disorder.

Although these clinical features typically resemble to those seen in already reported cases of Niemann pick disease and we didn't see any unusual/atypical feature in our case, however the purpose of discussing this case is to highlight the importance of evaluation of storage disorders in our population. In every child who presents with repeated infections, organomegaly and delayed milestones, we should consider evaluation of storage disorders.

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