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Niemann-Pick Disease: A case report

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Abstract

This case study's major objective to explore the limitations of treatment and management arisen of having **Niemann pick disease**, due to poor prognosis or lack of early diagnosis. Mrs. Pinki, 30 years old female patient from Chuadanga, with the complaint of upper abdominal swelling for 6 months, generalized weakness for last 3 months, anorexia & nausea for same duration, came to private chamber somewhere in Bangladesh, on 10th April 2023. She also complained generalized weakness for last 6 months associated with anorexia, nausea for same duration. Bone marrow examination confirmed the possible diagnosis of Niemann pick disease. Until her last follow up report, the patient was alive and clinically sound comparing to her previous condition. However, due to her lower socio-economic condition, she didn't continue the follow ups and could not able to do Sphingomyelinase enzyme level according to the physician's advice. Social awareness against consanguineous marriage & adding enzyme estimation facilities can a revolutionary change in reducing the mortality rate of this disease.

Keywords: Bone Marrow, Enzyme Estimation, Hepato-splenomegaly, Prognosis

Introduction

A rare neurodegenerative inherited disease, where fat metabolism of whole body is affected is called Niemann-pick disease. This life threating disease occurs due to mutations in either the NPC1 or NPC2 gene encompassing type A and type B as well as intermediate forms; Niemann–Pick disease type C (NP-C) including also type D. It is an autosomal recessive disorder, meaning that an individual needs to inherit a mutated gene from both parents to develop the condition. Affecting the brain, nerves, liver, spleen, bone marrow and, in severe cases, lungs, this kind of progressive malfunction can cause early death, most individuals dying between 10 and 25 years of age. Mainly it effects the children, however, it can also occur in adult age [1]. According to the previous studies, NP-C is estimated to occur in 1 case in every 120,000 live births [2].

There are a common scenarios of misdiagnosing NPC due to its undifferentiated clinical manifestations. The common complaints from the patients are, walking, swallowing, speaking, concentration, or memory. Overall, it distorts person's day to day normal life. Moreover, early death can happen in case of neurological dysfunctions [3].

There is no specific treatment of NPC. However, symptomatic treatment is suggested to inhibit the extreme progression of malfunctions by using multidisciplinary and multiprofessional teams of experts. As there is no specific curative treatment, and in cases where the diagnosis is delayed, the chosen treatment may be less effective or inappropriate for the advanced stage of the condition, potentially resulting in suboptimal outcomes. Hence, the prognosis of this disease is not satisfactory. The only motive of this supportive treatment is to improve the quality of life of the individual.

It's essential for patient communities like the International Niemann-Pick Disease Alliance (INPDA) to develop comprehensive disease management guidelines to enhance the care and support provided to individuals with Niemann-Pick disease (NPC). Creating standardized operating procedures can significantly improve the coordination and effectiveness of care across different regions and healthcare systems [4].

This case emphasizes the need to keep NPD in confirmed diagnosis of 30 years old female patient presenting with hepatosplenomegaly, associated with mild ascites.

Patient History

Mrs. Pinki, 30 years old female patient from Chuadanga, with the complaint of upper abdominal swelling for 6 months, generalized weakness for last 3 months, anorexia & nausea for same duration, came to private chamber somewhere in Bangladesh, on 10th April 2023. According to the statement of the patient, she was alright 6 months back. Since then, she noticed upper abdominal swelling associated with abdominal discomfort. He went to different type of physician; however, it remained undiagnosed. She took different types of medication like PPI, Domperidone, Timonium Methylsulphate and Metoclopramide, but there was no improvement found.

Moreover, she also complained generalized weakness for last 6 months associated with anorexia, nausea for same duration. However, there is no history of vomiting or hematemesis or per rectal bleeding or any blood transfusion. Patient had no recent history of travel to any hill tract. Her bowel & bladder habits were normal.

According to the patient's personal history, he took PPI, Domperidone, Tiemonium Methylsulphate & metoclopramide. Beside that, patient was fully immunized according to EPI schedule. He belonged to a middle-class family.

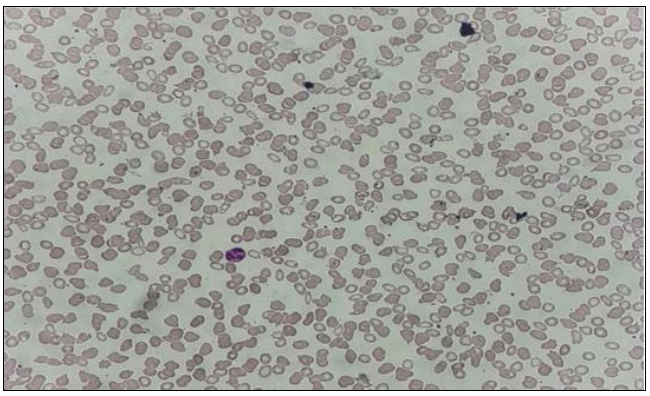
Clinical Presentation & Diagnosis

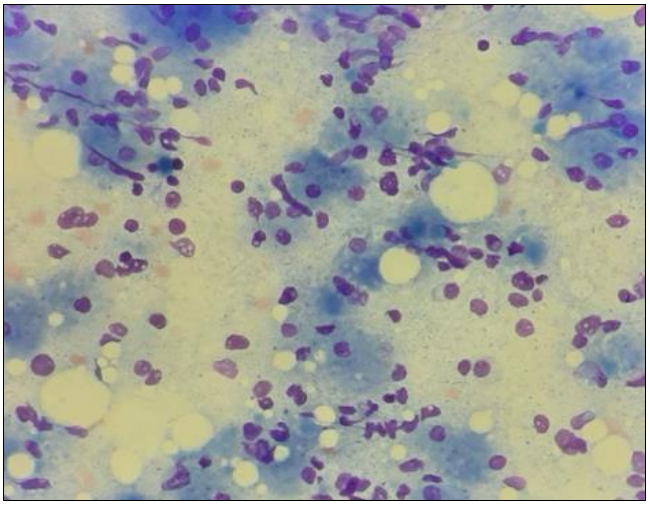
On general examination, the patient was ill-looking with average body built. No jaundice, cyanosis, clubbing, koilonychia, leukonychia or oedema was found. On systemic examination, upper abdomen was distended, umbilicus was centrally placed and inverted, no visible engorged vein peristalsis or scar mark was seen.

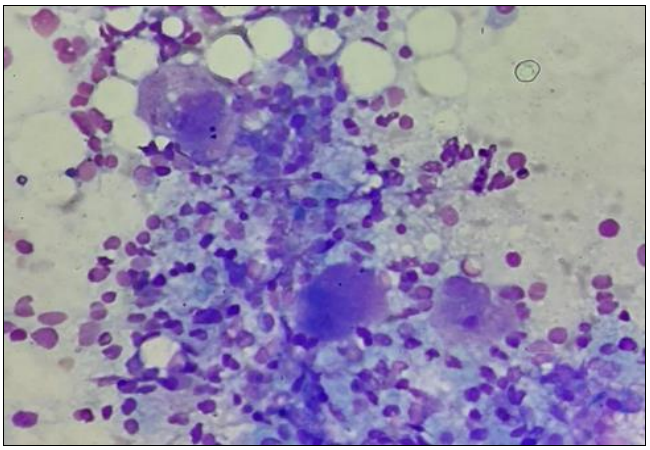
On systemic examination, liver was palpable 5xm from right costal margin in right mid clavicular line, firm in consistency, non-tender, smooth surface and splenic notch was present. Kidneys were not ballotable. There was no evidence of ascites.

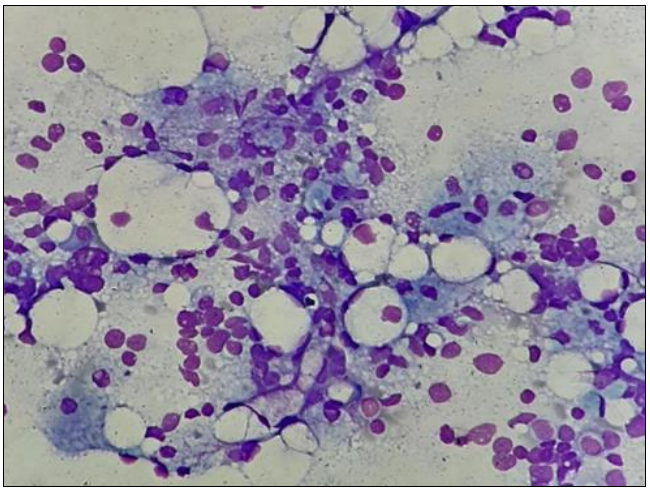
Patient was advised to do some regular routine investigations. There was strong evidence of iron deficiency anemia with leucopenia and thrombocytopenia in CBC report. Ultrasonogram report revealed hepato-splenomegaly with hick walled terminal ilium with mild ascites. Bone marrow examination confirmed the possible diagnosis of Niemann pick disease.

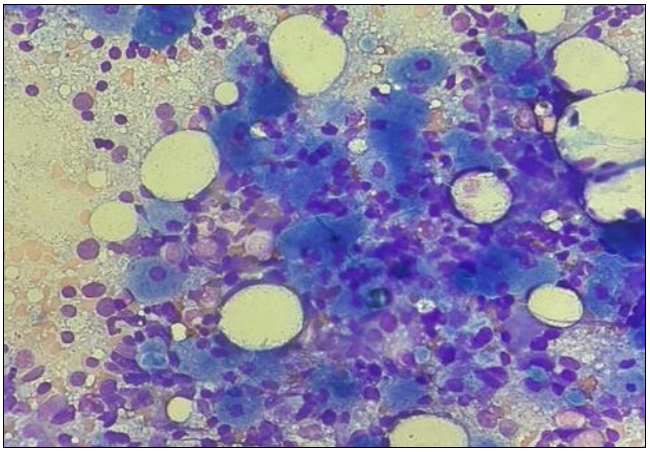
Fig 1: Niemann pick disease; a 30 years old female patient











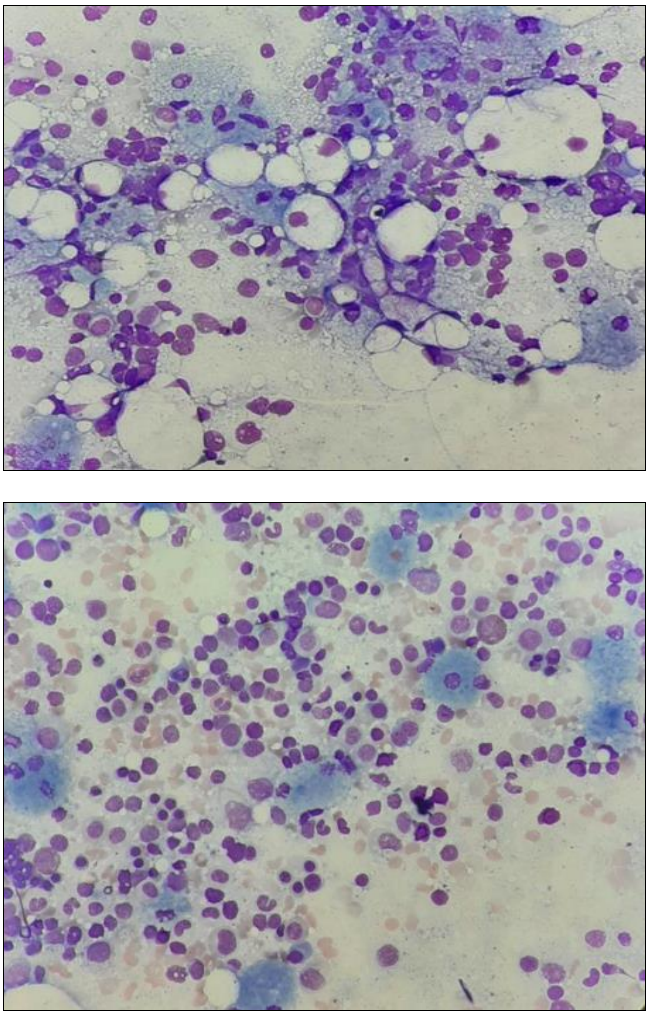


Fig 2: Bone marrow aspiration findings

Erythropoiesisis: Active with micro-normoblastic maturation.
Granulopoiesisis: Slightly depressed and maturing into segmented form.
Megakaryocytes: Are normal in number.
Histiocytes: *Plenty number of sea blue histiocytes are found with clusters. Fair number of foamy histiocytes are noted.*
Parasites are not seen.

Comment: **Features are suggestive of Storage disorder. (Most possibly Niemann pick disease).**

Treatment & Management

The patient was alive and clinically sound comparing to her previous condition until her last follow up report. However, due to her lower socio-economic condition, she didn't continue the follow ups and could not able to do Sphingomyelinase enzyme level according to the physician's advice. Moreover, the facility was not available in our country.

Discussion

Due to large range of symptoms, Niemann–Pick disease, a lysosomal storage disorder, is difficult to diagnose. So, the prognosis of this disease is not satisfactory. The manifestations depend on the age onset of the disease.

In South-East Asia, high prevalence of NP disease is found according to the previous clinical studies. In a previous case study, a 5 years old male child from Muzaffarnagar, UP state- presented with 15 days of fever, associated with respiratory distress, underwent routine examinations, however, storage disorder was confirmed by bone marrow examination. Unfortunately, he was died with severe illness at the 8th month of his age.

In another family history, 27-year-old female came for antenatal counseling in the fetal medicine clinic. According to her statement, two of her children expired in their early childhood due to seizures and hepatosplenomegaly. The enzyme analysis done in the second child was available and was consistent with NPD [6].

In Bangladesh, the prevalence of NP disease is still unknown. A 4 years boy presented with hepatosplenomegaly, repeated respiratory distress and growth failure, associated with Cherry red spot on his eye. Niemann Pick cell was confirmed by bone marrow examination. As the facility of enzyme estimation is still not available in Bangladesh. Patient was discharged by genetic counseling of parents [7].

Another case report was found in Department of Pediatric Neurology, Bangabandhu Sheikh Mujib medical University, Dhaka whereas, a 2.5 years female child who presented with developmental regression, recurrent seizures, failure to thrive and hepato-splenomegaly. Similar to other cases, Bone marrow (BM) aspiration was performed for confirmed diagnosis due to lack of facility of enzyme estimation. Follow up records revealed patient's improvement clinically in terms of nutritional status and there was no further deterioration of neurological symptoms and signs [5].

Among all the cases, we found that, usually patients were affected in an early age, however, in present case study, patient was female adult. On the other hand, one of the main obstacles for early diagnosis and satisfactory prognosis was, enzyme estimation facilities are not available in our country.

Conclusion

Though, in case of south-Asian epidemiology, Niemann pick disease is not common, the limitations of early diagnosis and treatment facilities should be more discussed in a broad spectrum due to the aggressiveness of this disease. Social awareness can be raised against consanguineous marriage to reduce the probabilities of this rare disease indeed.

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