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Case Report of Niemann-Pick Disease: Type -A

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Abstract: The Niemann-Pick disease is a rare lysosomal storage disease responsible for numerous cytological abnormalities involving various organs like liver, lungs and brain. The diagnosis requires enzymatic studies, which can be long and difficult in resource poor settings. In this context, the detection of cytological anomalies in blood and bone marrow smears, allowing a rapid screening, is an important step in the diagnostic approach. A review of English medical literature shows that 1,200 cases of NPA and NPB worldwide have been reported with the majority being Type B or an intermediate form. Since 1957, at least 27 cases of Niemann-Pick disease have been reported from India. The rarity of the Type A disease and difficulties in its diagnosis have prompted reporting of a new case of Niemann- pick disease that could be diagnosed on the basis of Clinical correlation and Liver Biopsy. A 11 months old child presented with feeding problems, abdominal distension. Bone marrow aspiration and Liver Biopsy was performed and revealed the presence of vacuolated cells. Diagnosis of type A Niemann-Pick disease was made by clinical correlation and Investigatory work up.
Keywords: Niemann- Pick disease, Bone marrow Aspiration, Liver Biopsy

INTRODUCTION

The Niemann-Pick disease is a rare lysosomal storage disease resulting in abnormal lipid metabolism causing harmful amounts of lipids [1] to accumulate in the spleen, liver, lungs, bone marrow, and brain [2]. Niemann-Pick disease types A and B are caused by an inherited deficiency of acid sphingomyelinase activity. The clinical phenotype ranges from a severe infantile form with neurologic degeneration resulting in death usually by 3 years of age (type A) to a later-onset non neurologic form (type B) that is compatible with survival into adulthood.

We report a case of 11 month old child with NPD who presented with splenohepatomegaly.

CASE REPORT

A 11 months old girl child, second of two children born of third degree consanguineous marriage hailing from Penuballi presented with feeding difficulties, fever, cough and progressive abdominal distension.

She had no history of jaundice or contact with tuberculosis. She had delayed motor milestones and was only able hold her head. Her elder brother was absolutely asymptomatic. She was a full term normal delivery with a birth weight of 2.2 kg without any antenatal or postnatal complications. On examination, she was malnourished (Fig. 1) with weight of 5.2 kg and height of 62 cms. She had pallor, generalized lymphadenopathy, fronto-parietal bossing, depressed nasal bridge

On systemic examination, she had splenohepatomegaly with a spleen palpable up to the umbilicus (14 cms) and liver span of 11 cms. Auscultation of chest revealed bilateral crepetations.

Fig. 1: Malnourished child

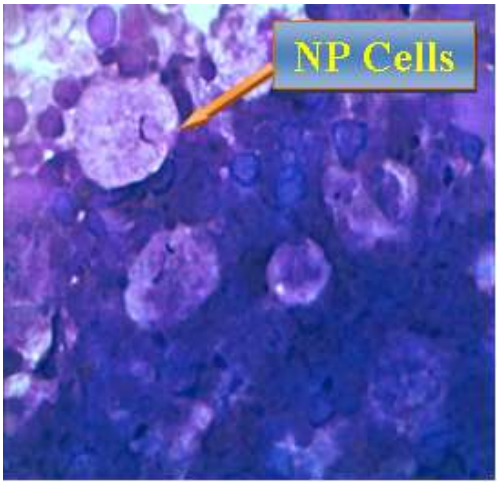

Fig. 2: Liver biopsy showing NP cells

Investigations showed anemia (Hemoglobin = 6.3% gm/dl, with neutropenia (WBC count = 9,200/cumm, 25% polymorphs, 72% lymphocytes, 2% monocytes and 1% eosinophils) and platelet count of 100,000/cumm) with a reticulocyte count of 1%. Her renal and liver function tests were normal. HIV by ELISA was negative and haemoglobin electrophoresis was normal.

Bone marrow aspiration was done and showed Niemann Pick's Cells and later confirmed by Liver biopsy (Fig. 2). Sphingomyelinase enzyme levels could not be done due to unaffordability. Child was discharged after 2week's and was on follow-up. Child developed severe respiratory problem after 2 months of discharge and died during transport to our hospital. Parents were advised for prenatal counselling for next pregnancy.

DISCUSSION

Niemann-Pick disease types A and B are autosomal recessive disorders, equally affecting males and females. The disease was first described by Albert Niemann [3, 4], in 1914. Ludwig Pick described the pathology of the disease essentially type B in a series of papers in the 1930s.

A review of English medical literature shows that 1,200 cases of NPA and NPB worldwide have been reported with the majority being Type B or an intermediate form. Since 1957, at least 27 cases of Niemann-Pick disease have been reported from India. Niemann pick disease (lipidosis) divided into 3 major types: Type A, B, and C. Nieman-Pick types A and B (NPA and NPB) are also referred to as, Acid Sphingomyelinase Deficiency (ASMD), and are caused by the deficiency of enzyme acid sphingomylineasse (ASM),resulting from mutation in SMPD gene [5]. This enzymatic defect leads to accumulation of sphingomyelin in the monocytes, reticulo endothelial system and in type A even in the central nervous system. Thus patients present with progressive lung disease, hepatosplenomegaly, short stature and pancytopenia.

Niemann Pick Disease type A is usually fatal [6] in childhood and patients may have prolonged jaundice at birth followed by hepatosplenomegaly, lymphadenopathy, psychomotor retardation [7] over a period of time as was seen in our patient. In later stages, spasticity and rigidity may be seen. Patients usually have a cherry-red spot on the fundus on ophthalmic examination.

Diagnosis of Niemann Pick Disease is by histopath finding of Niemann Pick cells on bone marrow examination. Diagnosis is confirmed by measurement of sphingomyelinase enzyme on cultured fibroblasts. Niemann Pick type A disease is usually fatal in childhood whereas prognosis for type B disease is variable. Treatment is supportive. Prenatal diagnosis with amniocentesis or chorionic villus sampling is available.

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