



Case Reports

Niemann Pick Disease: A Case Report

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Abstract

Niemann-Pick Disease is an autosomal recessive disorder of infancy characterized by failure to thrive, hepatosplenomegaly and neurodegenerative changes that leads to sphingomyelin and cholesterol storage within the lysosome.¹

Although the disease is common among Jewish individual but is rare in South Asian particularly in Bangladesh. So, we present the case as a rare incidence in our country.

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Introduction

Niemann-Pick Disease is an autosomal recessive disorder of infancy characterized by failure to thrive, hepatosplenomegaly and neurodegenerative changes that leads to sphingomyelin and cholesterol storage within the lysosome.¹

There are increased levels of sphingomyelin and cholesterol in bone marrow, liver, spleen and brain and the enzyme defect is sphingomyelinase deficiency. Failure to cleave phosphocholine from sphingomyelin results in the storage of sphingomyelin.²

Storage of cholesterol is not well understood, but there seems to be a close relationship between the metabolism of sphingomyelin and that of cholesterol.^{2,3}

Basically, two major classes exist. In one, there is a deficiency of sphingomyelinase activity (type A and B), and in other (type C and D)

sphingomyelinase activity is normal or only partially reduced in some cells.³

This disease begins at 3-4 months of age with feeding difficulties and failure to thrive. Neurologic function is gradually deteriorated and ultimately development is globally retarded.⁴

The diagnosis of this disease is based on hepatosplenomegaly, mental retardation, foam cells in the bone marrow, cherry red spot on the macula and sphingomyelinase deficiency in white blood cells, cultured fibroblast and other tissues as well.⁵

Although the disease is common among Jewish individual but is rare in South Asian particularly in Bangladesh. So, we present the case as a rare incidence in our country

Case report

R, a 19 months old child from Kumarpara, Ghoramara, Rajshahi came with history of

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apparently normal growth and development up to 5 months of age. There after the child developed gradual distension of abdomen, gradual regression of developmental milestone like disappearance of social smile, inability to sit, inability to hold neck and not able to recognize parents. All these regression started after 5 months of age, following an attack of pneumonia. Birth history was uneventful and the child was fully immunized.

On examination, the child had mild pallor and was afebrile, respiratory rate was 24/ min regular, Pulse rate 100/ min regular in volume and normal in rhythm.

Head circumference was 46 cm, (50th centile in NCHS); body weight 8.8 kg and height 78 cm. both were below (3rd centile in NCHS standard).

Liver was enlarged 10cm below costal margin. It was firm, smooth and non-tender.

Spleen was enlarged by 12 cm and it was non-tender.

All the deep reflexes were diminished and there was hypotonia in all four limbs. Sensory system could not be tested properly but pain sensation was preserved. Blood examination showed: reduced hemoglobin

Hb%- 8.9 gm, TLC-3000/cm, DLC-N-65%, L-35%, Reticulocyte count- 1.3%, Blood urea-14mg%, Serum total protein-5.9 gm%, Albumin-3.8 gm%, Globulin 2.1 gm%, Serum electrolytes- Na^+ -133 mmol/L, K^+ -3.9 mmol/L, Prothrombin time- 13.1 sec.

X-ray chest showed nothing abnormality. USG reflected hepatosplenomegaly with normal echotexture without ascites.

Fundal examination revealed cherry red spot in the macula in both eyes. Bone marrow showed infiltration of numerous foamy cells in the macrophage. There was also normoblastic erythroid hyperplasia.

Discussion

Niemann Pick disease has not been reported to the knowledge of the author in Bangladesh. It

has been described as a very rare disease in childhood.^{1,3} The most common from of Niemann pick disease (Type A), i.e., Acute neuropathic form present in the first months of life with marked hepatosplenomegaly, failure to thrive, psychomotor retardation, half of the patients have a cherry red spot in the macula, there is wide spread infiltration of foam cells in the lungs, bone marrow and most patient die in the first year of life.^{3,5} Our case coincides with the type A acute neuropathic form of Niemann pick disease and ultimately patient died at the age of 22 months old.

Other Variant of Niemann- Pick disease-particularly type B (chronic non neuropathic from) type C (sub acute neuropathic variant), type D (Nova Scotian variant) present in early childhood with less severe symptoms than in Niemann- Pick disease (type A).

Diagnosis of these diseases depends on sphingomyelinase deficiency in leukocytes or cultured fibroblasts. But the deficiency is more marked in the type A variant. We could not determine the enzyme analysis due to lack of facilities.

There are various types of lipid storage disease. Among them Juvenile Tay-Sachs disease and Hurler's syndrome have more or less similar presentation with that of Niemann-Pick disease. But in former motor weakness, delayed development, poor co-ordination and subtle response to sharp sound begin at 3-6 months of age although on fundoscopy a cherry red spot in the macular region, but no visceromegaly and deficiency of hexosaminidase enzyme, in the WBC and cultured fibroblast.⁷

In Hurler's syndrome- manifests at 6-12 months of age affected infants are large weight dolicho-cephalic head, mucoid rhinorrhoea, hepatosplenomegaly, coarse facies, corneal clouding, claw hand deformity contracture at the elbow, shoulder, hip and knee, gibbus deformity with a deficiency of α -L- iduronidase in the WBC and cultured fibroblast.⁸

Another infantile Gaucher disease-presentation similar to Niemann-Pick disease which occurs within the first few months of life-

hepatosplenomegaly, strabismus, swallowing difficulty, opisthotonos, laryngeal spasm and Gaucher cells in the bone marrow due to deficiency of galactosidase activity could easily be differentiated from Niemann-Pick disease.

In Niemann-Pick disease the extent of abnormality may vary considerably even between affected siblings, but our case had all the cardinal features like presentation in early months of life, huge hepatosplenomegaly, mental retardation, cherry red spot in the macula and foam cells in the bone marrow.

Although bone marrow transplantation tried in some centers all over the world but the results were unsatisfactory. So prenatal diagnosis and carrier detection is necessary for the control of this disease.



Fig-1: Marked hepatosplenomegaly in a patient of Niemann-Pick disease.

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