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Case Report

A Rare Case of Infantile Type of Sandhoff Disease

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ABSTRACT

We report a rare case of inherited metabolic disease wherein one and half year old girl child born to a consanguinously married couple, presented to emergency ward with a history of one episode of generalized tonic clonic convulsion lasting for about one hour, followed by post ictal drowsiness. On examination child was conscious, playful, not following objects. There was global developmental delay. There was generalized hypotonia, exaggerated deep tendon reflexes, and reduced power in all the four limbs. Fundoscopic examination showed bilateral cherry red spots. Hematological examination was normal. On further evaluation for inborn errors of metabolism, total serum hexosaminidase was less than 100 nmol/hr/ml suggestive of Sandhoff disease. Anticonvulsant treatment and supportive management was given to the child.

Key words: GM2 gangliosidosis; Cherry red spot; Sandhoff disease; Developmental delay.

INTRODUCTION

Sandhoff disease is a rare autosomal recessive inherited metabolic disorder caused by mutation in the HEXB gene on chromosome 5q13. [1] It results from defects in the beta chain of beta hexosaminidase. [2] Incidence of the disease is 1 in 384000. [3] There are three different types of Sandhoff disease, classic infantile, juvenile, and adult late onset. [4] As there is no treatment and antenatal diagnosis in risky parents is the only means of preventing the disease, hence we are reporting the case.

CASE REPORT

One and half year old female child born to a consanguineously married couple presented to emergency ward with a history of one episode of convulsion. It was generalized tonic clonic type of convulsion with uprolling of eyeballs, presented to us in state of post ictal drowsiness. The child was born in hospital and there was history of weak cry at the time of birth, which did not need any admission during neonatal period. There was also delay in attaining developmental milestones. Child did not attain neck holding or rolling over, does not reach for objects and does not recognize mother. There was delay in language development. At the age of one year child also had similar episode of convulsion, admitted in outside hospital and was started on sodium valproate since then. On examination child was conscious, playful, startle response for loud sound was present

but was not following objects. There was mild hepatosplenomegaly and bony changes (fig 1). Fundoscopic examination showed cherry red spots in both the eyes. Hematological examination showed following results Hb 11.5gm%, TC 11,600/cmm, platelet count 3.43lac/cmm, PCV 29.6%, CRP negative. RBS 119mg%, Serum Na 138 meq/l, serum K 6.3meq/l, Cl 109meq/l, serum Ca 8.2mg%, blood urea 33mg%, Serum creatinine 0.9% ,total serum bilirubin 0.3mg%, direct 0.1mg%. SGOT 177.6 Iu/L, SGPT 139.6Iu/L, alkaline phosphatase 171.6 Iu/L, total protein 6.5gm%, serum albumin 4.1gm% gamma GT 70.5Iu/L. Total Hexosaminidase level was < 100nmol/hr/ml (normal 333-1500) which confirmed the diagnosis of Sandhoff disease. Child was treated with anticonvulsants and supportive management was given as per the unit protocol.

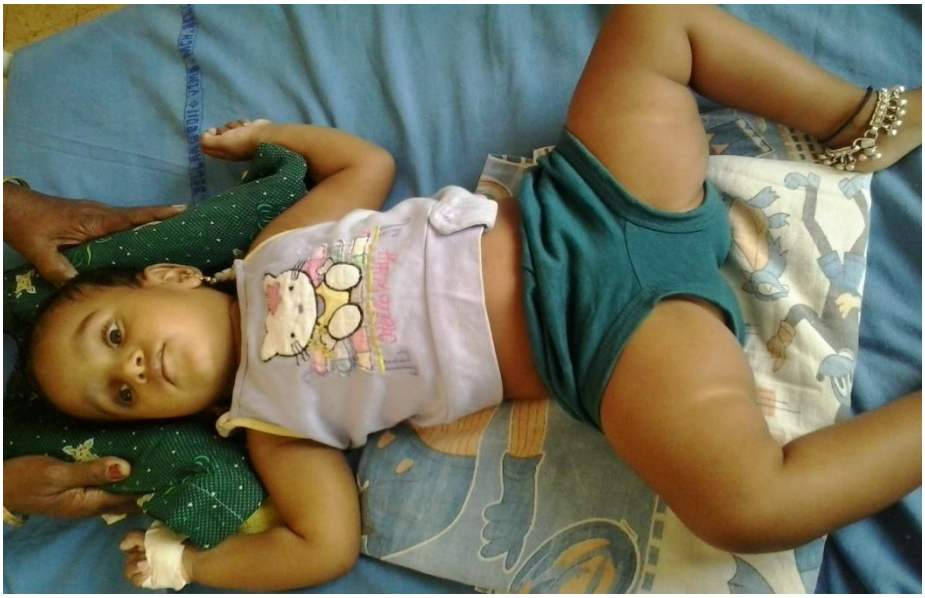

Fig 1. Showing hypotonia and loss of visual fixation.

DISCUSSION

Sandhoff disease is a rare and severe lysosomal storage disorder representing 7% of GM2 gangliosidoses. [5] patients with the infantile Sandhoff disease will have increased startle reaction and macular pallor and retinal cherry red spots., decreased eye contact, hepatosplenomegaly, cardiac involvement and mild bony abnormality. [6] Hypotonia and acoustic hypersensitivity were among the most common early symptoms, followed by hypertonia/spasticity and subsequent loss of hearing. Seizures were a late but common symptom. [7] The diagnosis is usually suspected in an infant with neurologic features and cherry red spots. Definitive diagnosis is by determination of the levels of hexosaminidases. [6] Bilateral thalamic involvement has been suggested as a diagnostic marker of Sandhoff disease. [5] Early seizures seemed to be a marker of disease severity, or at least worse motor developmental outcomes. [7] No correlation was evident between the severity of the central nervous system imaging and clinical picture. [8] The classic infantile form is known to lead to death between 3 and 5 years of life, but the rate of functional decline remains poorly defined. [9] Treatment

is supportive. Prenatal diagnosis is available by the detection of N-acetylglucosaminyl oligosaccharides in amniotic fluid. [10]

CONCLUSION

Infantile GM2 gangliosidosis is one of the inherited neurologic disorders with relentless progression. Delay in developmental mile stones and seizures will pose lot of difficulties in managing the case. As supportive treatment is the only option, timely diagnosis during antenatal period in high risk parents will help in reducing the disease burden.

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