Research Paper Medical Science

Late Infantile Onset Gangliosidosis in A 1 Year Old Girl- A Rare Case Report

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KEYWORDS : Tea, Auction, Marketing, Export, Consumer, Economy.

INTRODUCTION :

GM1 gangliosidosis is a rare genetic disorder caused by mutations in GLB1 gene leading to the deficiency of enzyme beta galactosidase. [1]. The clinical manifestations are varied due to accumulation of ganglioside in the lysosomes. It can be divided into three types depending on the age of onset. Type 1 is an infantile form, which presents between birth and 6 months of life. Type 2 is the late infantile form, the onset varies between 6 months and 3 years of age, the clinical features of which include neurological deterioration, and cerebellar and extrapyramidal symptoms. Organomegaly, cherry red spot and skeletal changes are usually not observed in this form. Type 3 (chronic/adult form) presents between 3 and 30 years assay or mutation. The disorder can be diagnosed in several ways, including lysosomal enzyme assay of low beta-galactosidase activity in peripheral leukocytes or cultured skin fibroblast, detection of abnormal urinary oligosaccharide excretion and rectal biopsy[2]. Prenatal diagnosis by measurement of enzyme activity in amniotic fluid and cultivated amniotic fluid cells has also been extablished[3]. We present a child with late infantile form of GM1 gangliosidosis.

CASE REPORT :

A one -year-old girl was brought with the complaint of regression of developmental milestones. She gained normal developmental milestones till the age of 7 months after which she gradually lost ability to sit, neck control, and social smile. There is no history of seizures. There is no history of any consanguinity in the family. Antenatal period is uneventful. She is born by normal vaginal delivery with no history of any postnatal or neonatal complications. There is a history of similarly affected elder female sibling who died at 3 years of age. On examination, weight, length and head circumference were below -2SD. There is spasticity in the upper and lower limbs and the deep tendon reflexes are absent. There is no startle response, no fixation to light, no nystagmus. Hepatospenomegaly is present. Fundus examination showed bilateral optic atrophy with cherry red spots (figure 1). MRI (magnetic resonance imaging) brain showed Axial T1-weighted image and T2-weighted image at the level of thalamus show diffuse dysmyelination of white matter with bilaterally symmetric thalamic signal change, which appear hyperintense on T1-weighted and hypointense on weighted images (figure 2)

Figure 1: Bilateral optic atrophy with cherry red spots.

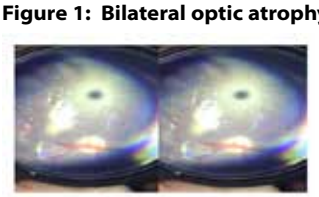


Figure 2: MRI findings

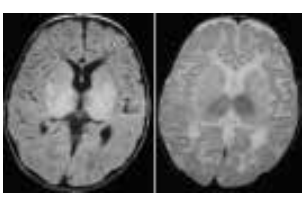
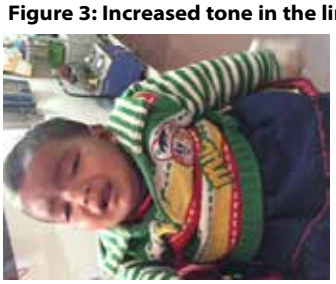


Figure 3: Increased tone in the limbs.



DISCUSSION :

Infantile GM1gangliosidosis, the most common and severe form, is characterized by facial and skeletal abnormalities and neurological deterioration before the age of 6 months. Death usually occurs before the second birthday.[1] This disorder can be diagnosed by several ways, including lysosomal enzyme assay of low β-galactosidase activity in peripheral blood leukocytes or cultured skin fibroblasts, detection of abnormal urinary oligosaccharide excretion, and rectal biopsy .White matter abnormality in late infantile GM1 gangliosidosis have rarely been reported previously [4-6]. Moreover, optic atrophy is a rare eye manifestation seen in this disorder . Neuroimaging findings in late infantile GM1 gangliosidosis have been rarely reported. In one case , initial thalamic hyperdensity was found on CT-scans and hypointense signal of the thalami was seen on T2 weighted MR images at a later stage[7]. Persistent delay in white matter myelination on serial MR studies has been described in another case[8]. Our case report highlights the MRI imaging and eye findings in late infantile GM1 gangliosidosis which have been rarely reported previously. For more precision, enzyme asssays were send on follow up which showed results of decreased activity of beta galactosidase in the given blood sample. The patient was put on a regular follow up with multidisciplinary involvement of various specialists and prognosis and life expectancy was explained to the parents.

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