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Case Report

Inclusion Cell Disease - A Rare Cause of Megalocornea with Corneal Edema

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Abstract *Inclusion cell disease (I cell disease), also known as mucolipidosis type II, is a rare congenital metabolic storage disorder which seemingly occupies an intermediate position between mucopolysaccharidosis and sphingolipidosis.[1] We are hereby presenting a case of I cell disease that presented with megalocornea and corneal edema/corneal clouding.*

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Keywords: *Inclusion cell disease, mucolipidosis, sphingolipidosis, megalocornea, corneal edema, corneal clouding.*

Introduction

There are multiple causes of megalocornea and corneal edema in infantile age group. The common causes include birth trauma, Peter's anomaly, limbal dermoid, sclerocornea, congenital hereditary endothelial dystrophy (CHED), mucopolysaccharidosis and infective/inflammatory process. It is important to consider congenital error of metabolism including mucopolysaccharidosis as an etiology for corneal edema/corneal clouding and megalocornea because these cases are rare and can be easily missed out if not evaluated properly.

Case report

A 11 month old male child presented with complaints of reduced weight gain, failure to thrive, recurrent respiratory infections and diarrhea. On clinical examination the patient showed coarse facies, flat face, prominent bulging eyes, large tongue, enlarged gums (Figure 1). Perinatal history was normal term vaginal delivery, and he was the first child of non-consanguineous marriage. Ophthalmological examination revealed megalocornea (horizontal corneal diameter of 16 mm – Figure 1) with corneal edema/corneal clouding. Anterior chamber was deep. No cherry red spots were seen on fundoscopic examination. Intra ocular pressure was within normal limits. On Gonioscopy, angles were open. On indirect ophthalmoscopy, the optic disc was of normal size, shape & colour. The optic cup was within normal limit. The neuroretinal rim was healthy. Retinal artery:vein ratio was maintained. The macula was healthy and the foveal reflex was present.

MRI Brain was done which revealed T2 hyperintense signal involving cerebral white matter on either side uniformly involving both subcortical and periventricular white matter without abnormal T1 hypointense signal suggestive of Hypomyelination MRI is (Figure 2). T2 hypointense signal was seen in bilateral thalami (Figure 2). Therefore possibility of lysosomal storage disorder was considered.

CT skull revealed premature fusion of cranial sutures suggestive of craniosynostosis (Figure 2). No anterior vertebral body beaking was noted on lateral X-Ray of dorso-lumbar spine. X-Ray pelvis with femur revealed osteopenia with short thick femur (Figure 3). Ultrasonography of abdomen showed mild hepatosplenomegaly with liver

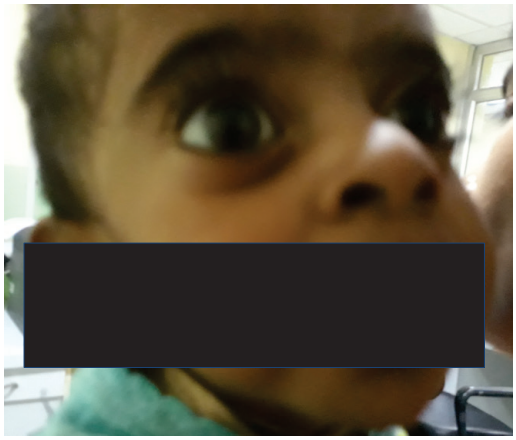

Figure 1: *Clinical photograph of patient showing bulging eyes, megalocornea with flat and coarse facies.*

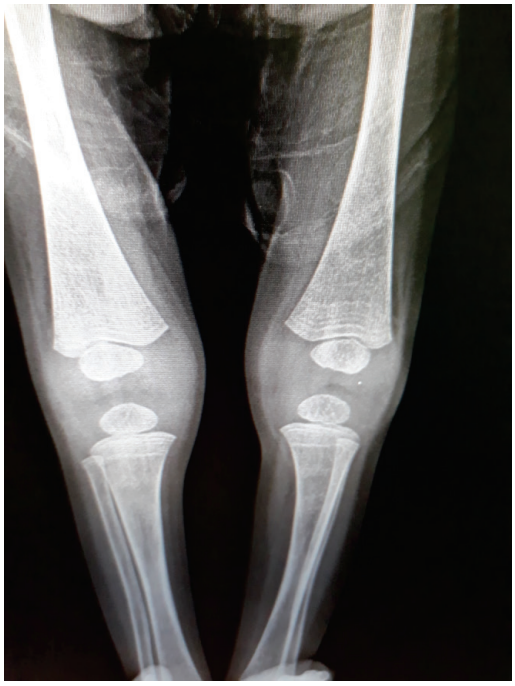

Figure 2: *MRI bilateral knee joint AP view showing mild osteopenia with short thick femur.*

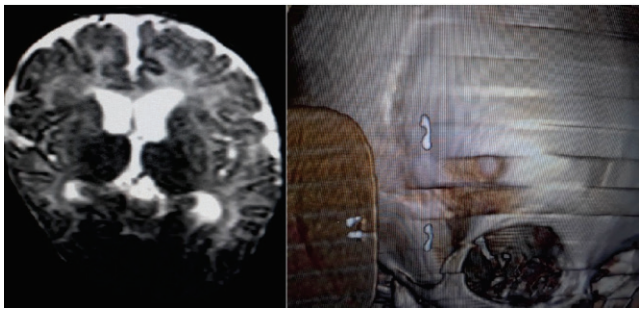

Figure 3: *X-ray Brain axial T2W sequence showing hyperintense signal in cerebral white matter with bilateral T2 hypointense thalami. Volume rendered reformated Computed Tomography image of skull showing fusion of coronal suture and craniosynostosis*

function test showing mildly elevated bilirubin and liver enzymes with changes of mild cholestasis. Thus, a high suspicion of Inclusion cell disease was made which was confirmed on lysosomal enzyme activity.

Discussion

Mucolipidosis is a rare congenital error of metabolism that has an intermediate position between mucopolysaccharidosis and sphingolipidosis.[1] The significant ocular manifestation of mucolipidosis are common to both mucopolysaccharidosis and sphingolipidosis. These ocular manifestation are variable and include corneal clouding in generalized gangliosidosis, mucolipidosis type I, II, III and macular cherry red spots in generalized gangliolipidosis, mucolipidosis type I and Farber's disease. Mucolipidosis type II, the I-cell variant, is a rare disorder which was first described in 1967 by Leroy and De Mars,[2] who found coarse cytoplasmic inclusions in cultured fibroblasts (which they designated I-cells or inclusion cells) from two children with this disease. It is transmitted as an autosomal recessive trait which is present at birth and is associated with severe psychomotor retardation, early cessation of growth, Hurler-like facies with characteristic gingival hyperplasia, and extreme skeletal dysplasia. Additional clinical signs include mild hepatic enlargement restricted joint mobility, and unusually tight skin.[3] Luchsinger et al[4] have described corneal opacities in case of probable Mucolipidosis type II with absent macular cherry-red spot. Early eye involvement has also been described with I-cell disease in other studies. Of 35 patients with I-cell Disease studied by Libert J et al, the most frequent ocular finding was corneal clouding (14 cases), glaucoma (2 cases), or megalocornea (2 cases).[5] There were no descriptions of macular cherry-red spots, a frequent later finding in several of the Lysosomal storage disorders. Ultrastructural study of the eyes in 7 affected patients revealed changes in the conjunctival, corneal, scleral, and uveal fibroblasts, whereas other cells were not damaged.

Neonatal hepatosplenomegaly has also been described in cases of I-cell disease and multiple sulfatase deficiency.[6-8] I-cell disease has been described together with craniosynostosis in the neonate in a few cases. Prenatal development of short femurs has also been seen in I-cell disease and must be differentiated from neonatal osteogenesis imperfecta.[9] I-cell disease should be part of the differential diagnosis of significant craniosynostosis in the neonate or prenatal diagnosis of short femurs, especially if coarse facies is seen postnatally.[10] Thus in cases of infants presenting with megalocornea & corneal edema, the possibility of I- cell disease should be kept in mind if associated coarse facies are present & these cases should be further evaluated with MRI brain, ultrasound abdomen & skeletal survey.

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