Letters to the Editor

Basal Ganglia Changes: A Diagnostic Clue to Sandhoff Disease

Sandhoff disease is a rare autosomal recessive metabolic disorder of GM_2 gangliosides. Recently, a 10-month-old female child of nonconsanguineous marriage presented with developmental delay. She had attained social smile and approach to objects. There was no head control. She was not able to recognize her parents. There were no seizures. Vision and hearing were normal. However parents denied any deterioration in the neurological state. Growth parameters including head circumference were within normal limits. She had mild hypotonia. Deep tendon reflexes were normal and plantar responses were flexor bilaterally. She had bilateral cherry red spots in the fundus. Enzyme assay from two separate laboratories showed marked reduction of hexosaminidase A and B levels in the serum. Magnetic resonance imaging (MRI) showed unmyelinated large parts of white matter of centrum semiovale. Striking putaminal hyperintensity was seen on T_2 weighted images bilaterally (*Fig. 1*). Thalami also showed low signal on T_2 weighted images bilaterally.

Studies on magnetic resonance imaging in Sandhoff disease are scant in literature. However as early as in 1993, Caliskan, *et al.*(1) have suggested bilateral thalamic hyperdensity on computed tomography as a diagnostic marker of Sandhoff disease. Several other reports have shown involvement of thalamus, basal ganglia (caudate, putamen, globus pallidus) and cerebellum in this condition with rare involvement of other parts of brain(2-5). A review of these works suggests basal ganglia are more consistently involved than the other regions of the brain. Based on findings in three patients with GM_2 gangliosidosis Grosso, *et al*. have suggested that MRI pattern peculiar to GM_2 gangliosidosis can be defined(4). There have been a few attempts at clinical correlation with neuroimaging(3,4).

Our case shows further evidence to basal ganglia involvement in Sandhoff disease. We alert our fellow physicians involved in evaluation of neurometabolic diseases to look for these findings in more cases and perform

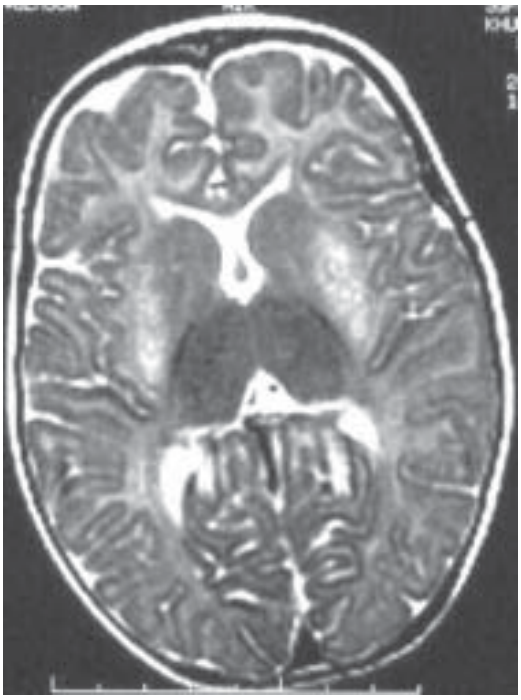

Fig.1. Bilateral putaminal hyperintensity on T_2 weighted images

enzyme assay when such lesions are noted on MRI. Any neuroimaging clue for the diagnosis of neurometabolic disorders are of paramount importance as diagnosis based on clinical information is very difficult and lack of access to biochemical assays in Indian scenario.

Acknowledgements

The authors thank Dr. Rajendra V. Phadke, Professor of Radiodiagnosis, Sanjay Gandhi Postgraduate Institute of Medical Sciences for the radiological evaluation.

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Chiari Malformation Type II with Vanishing Cerebellum

An eight-month-old baby, the second of two siblings born to non-consanguinous parents, presented with delayed milestones and a history of a lumbosacral swelling which was operated in the early neonatal period.

MR imaging revealed a small posterior fossa occupied by the occipital lobes and a profoundly small cerebellum (*Fig 1*). The tentorial incisura was heart-shaped. Sagittal sections demonstrated hypoplastic cord-like cerebellar tonsils herniating through the foramen magnum into the upper cervical canal and an elongated poorly-formed fourth ventricle (*Fig. 2*). Other findings were a small-sized pons with loss of normal pontine prominence, caudal elongation of the medulla and beaked tectal plate. The torcular heterophili was low-placed and supratentorial hydrocephalus was present. The massa intermedia was absent and the falx was hypoplastic with consequent interdigitations of gyri.

MR images of the lumbosacral spine revealed dysraphism and lumbar meningomyelocele with tethering of the cord.

In 1891, Hans Chiari first described an anomaly encompassing elongated peg-like cerebellar tonsils displaced into the upper cervical canal through the foramen magnum to be later known as the Chiari type I malformation. Chiari type II anomaly includes herniation of the medulla, fourth ventricle and cerebellar vermis through the foramen magnum. Chiari III combines features of