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IMAGES IN PEDIATRIC SCIENCES

Hallervorden spatz syndrome: eye of the tiger

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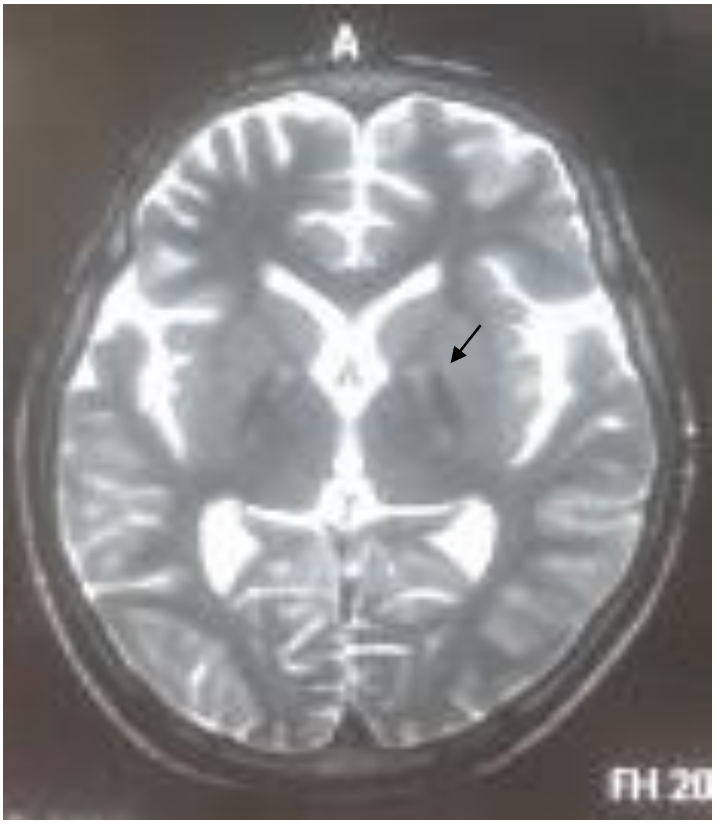

Figure 1. T2W axial MRI images show the classic "eye-of-the-tiger" sign, with marked hypointensity in the medial globus pallidi with a small area of central hyperintensity (arrow).

11 years old boy came to our department with abnormal posturing of head and limbs and difficulty in walking since last 4 years. Parents noticed that since 5 years of age he had history of frequent falls which later progressed to stiffness and abnormal posturing in lower limbs progressing to the upper limbs. There was history of seizures (generalized tonic clonic) since last 3 days . For last 1 year he has developed contractures at ankle, knee, elbow and wrist joint. Patient also developed twitching movements of face and drooling of saliva. He had progressive decline in scholastic performance and cognitive functions. Presently child was completely bedridden and produces some sounds only. He was constantly crying, not able to sleep , feed himself as well as swallow food or tell about urine and stool.

Patient was born vaginally , immediately cried at birth with no history of neonatal jaundice. His pre and post birth history was uneventful and attained developmental milestones at appropriate age till 5 yrs of age. Family history revealed same complaints in 2 cousin siblings i.e. children of maternal aunt and paternal uncle. On examination of index case child was thin built, malnourished with healed scar marks present on forehead and was irritable. There was generalised hypertonia with lead pipe rigidity and marked dystonic posturing which increased on activity with presence of facial dystonia and oromotor dyskinesia. Spasticity was noticed in tendoachilles, hamstrings and adductor muscles. There was hyperreflexia with bilateral extensor plantar response. Speech disturbances including dysarthria and drooling was also present. Cranial nerves and sensory system examination was normal.

The two cousins, 8 years female and 10 years male child in family also had h/o recurrent falls and injuries on forehead but severity was less . Onset was at 5-6 years of age in all three of them. There were relatively infrequent episodes of dystonic posturing with slow gait due to presence of contractures. Their speech had minimal spontaneity and was dysarthric and sluggish. Female child was less affected. No history of seizure and cognitive dysfunction in both of them.

Serum electrolytes, iron, copper, and ceruloplasmin levels were normal. MRI brain revealed area of hyperintensity within a region of hypointensity in medial globus pallidus bilaterally on T2 images “eye of the tiger” pattern (Figure1). Genetic studies could not be done due to financial reasons. Peripheral smear did not show acanthocytosis.

Eye examination did not reveal Kayser-Fleischer ring and retinitis pigmentosa.

Hallorverden – Spatz syndrome encompasses a group of rare neurodegenerative disorders with an incidence of 1-3/1,000,000 based upon observed cases in population. Clinical features include early onset of progressive dystonia and intellectual impairment. Dystonia, dysarthria, rigidity and choreoathetosis is seen in 98% of cases, cortical tract sigs in 25%, cognitive decline in 29%, optic atrophy in 3% and acanthocytosis in 3% cases. Hayflick et al. (1) classified the neurodegeneration with brain iron accumulation (2) in different groups on basis of age of onset and gene defect. The classical form, with pantothenate kinase 2 mutation, is characterized by early onset, rapid progression, and presence of the typical eye-of-the-tiger sign.

The eye-of-the-tiger sign on the MRI scan and clinical findings has contributed to a diagnosis of Hallervorden-Spatz disease for this patient. The typical MRI findings include bilaterally symmetrical, hyperintense signal changes in the anterior medial globus pallidus, with surrounding hypointensity in the globus pallidus, on T2-weighted images (3,4). The ‘eye of tiger’ sign is fairly specific and hence can be used to identify patients for pantothenate kinase 2 genetic testing and has accurately identified pre-symptomatic siblings of affected children.

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