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CASE REPORT

A Case Report of Hallervorden-Spatz Disease with Classical Magnetic Resonance Imaging Feature

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Abstract

Hallervorden-Spatz disease is a rare neurometabolic disorder characterized by iron accumulation in the brain, manifesting with extrapyramidal symptoms, psychosis, dementia, and retinal degeneration. This report presents a case exhibiting dystonia, retinitis pigmentosa, and psychotic symptoms, accompanied by classical magnetic resonance imaging (MRI) feature. MRI of brain of two asymptomatic siblings of index case also demonstrated the characteristic "eye of the tiger" sign.

Key words: *Hallervorden-Spatz disease, Eye of the tiger sign.*

Introduction

Hallervorden-Sptaz disease, also referred to as pantothenate kinase-associated neurodegeneration (PKAN), is a rare neurological disorder characterized by extrapyramidal symptoms, cognitive decline, and retinitis pigmentosa. This condition has been observed in both sporadic cases and familial occurrences. We present a case study involving three siblings within a single family who were diagnosed with this disease, exhibiting the characteristic MRI findings.

Case report

A 16-year-old male, the second offspring of a non-consanguineous marriage, presented with abnormal posture of the neck and right hand, as well as difficulty in ambulation for one year. His symptoms commenced 5 years prior, manifesting as irrelevant speech, self-muttering, and occasional episodes of aggression. Subsequently, he developed progressive vision loss, initially occurring nocturnally. He reported no history of ocular pain, erythema, photophobia, or visual phenomena such as floaters or flashes. The patient had a history of normal vaginal delivery with no reported perinatal asphyxia or neonatal jaundice. His developmental milestones were within normal limits; however, his scholastic performance began to decline gradually in grade 5, ultimately leading to the discontinuation of his education. The patient had two brothers, both of whom appeared to be in good health. No other family members, to the extent known, were afflicted with a similar condition. The patient's speech was characterized by low volume and sluggishness, with minimal spontaneity. No Kayser-Fleischer ring was observed on ocular examination. Fundoscopic examination revealed features consistent with retinitis pigmentosa, including numerous scattered yellow-white spots and bony spicule pigmentary lesions in the peripheral retina bilaterally. The patient exhibited generalized hypertonia and marked dystonic posture of the neck, as well as a flexor posture of the right hand. All reflexes were

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exaggerated with bilateral plantar extensor responses. Laboratory tests, including serum electrolytes, iron, copper, and ceruloplasmin levels, were within normal limits. Peripheral blood film was unremarkable, with no evidence of acanthocytosis. Magnetic resonance imaging of the brain revealed areas of hyperintensity within regions of hypointensity in the medial globus pallidus bilaterally on T2-weighted images, consistent with the "eye of the tiger" pattern (Figure 1). Genetic studies were not performed. Fundus photography confirmed features of retinitis pigmentosa.

While the patient's two siblings exhibited no symptoms, brain MRI scans revealed distinct findings. The older brother's scan demonstrated the characteristic "eye of the tiger" sign (Figure 2a). Conversely, the younger brother's brain MRI displayed initial changes, characterized by a hypointense signal in the medial globus pallidus, with a hyperintense area beginning to manifest within it (Figure 2b).

Based on the clinical presentation and magnetic resonance imaging (MRI) findings, a diagnosis of Hallervorden-Spatz Disease was established. Genetic

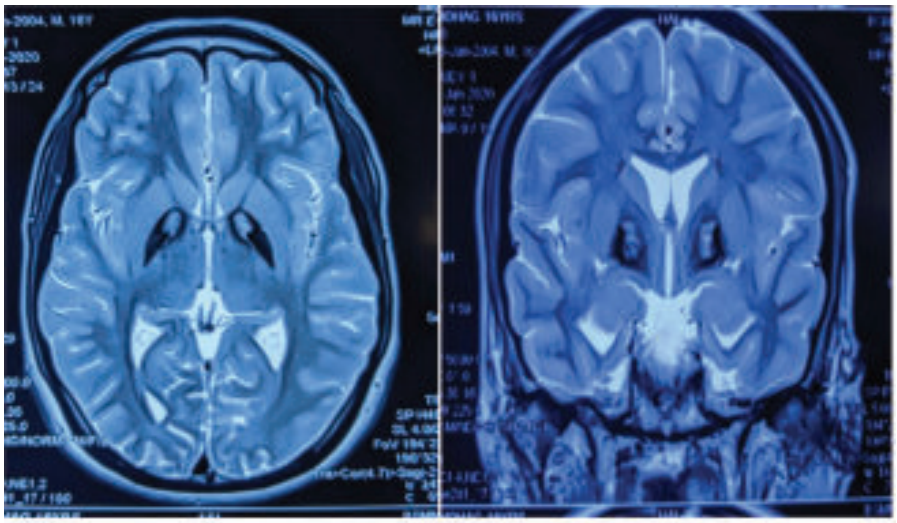

Figure 1: *T2W axial and coronal MRI images show the classic "eye of the tiger"sign, with marked hypointensity in the medial globus pallidus with small area of central hyperintensity.*

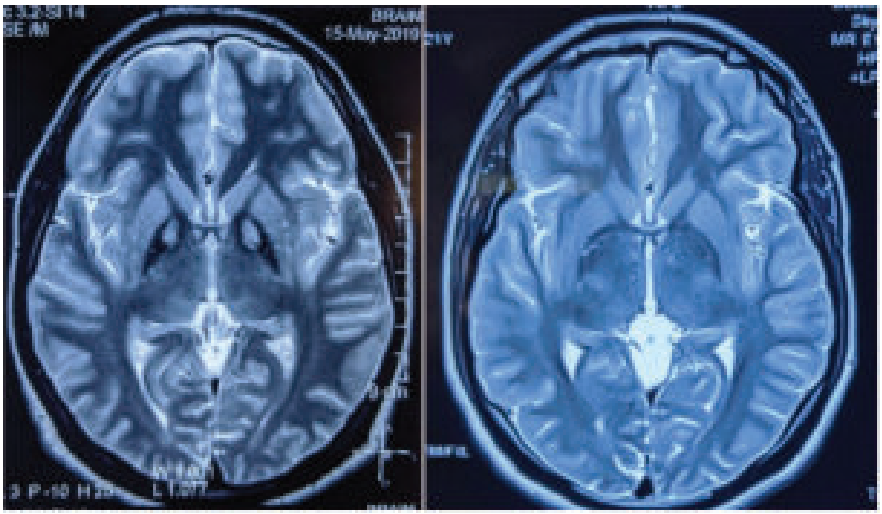

Figure 2 a-b: *T2W axial MRI of two asympotomatic brothers of index case showing (a) classical "eye of the tiger" sign in elder brother and (b) medial globus pallidus hypointensity but within it hyperintensity is less marked in MRI of younger brother.*

study could not possible due to patient financial constrain. The patient was administered symptomatic treatment. Additionally, physiotherapy was initiated.

DiscussionHallervorden Spatz disease is a rare neurodegenerative disorder also known as Pantothenate Kinase-Associated Neurodegenatation (PKAN). It was first described by Hallervorden and Sptaz in 19221. It may occure as familial (autosomal recessive) or sporadic disorder[2].

The familial form associated with mutations in pantothenate kinase gene (PKAN2) on chromosome 20p12.3-132 .

Pantothenate kinase is essential for phosphorylation of pantothenic acid that lead to under utilization of cystine and excess cystine causes chelation of iron leading to free toxic radicals production. Number of pantothenate kinase receptor are more in basal ganglia region that lead to preferential involvement of this region3.

Clinical features are early onset progressive dystonia, parkinsonism, choreoathetosis and intellectual impairment[4].

“Eye of the tiger” sign charactaristic feature of MRI of brain is corresponds to the pathological finding. T2 hypointensity in the anterior medial globus pallidus due to iron deposition and central hyperintensity due to gliosis and spongiosis[5].

Other differential diagnosis of “eye of the tiger” sign on MRI in the basal ganglia are early onset levodopa responsive parkinsonism, aceruloplasminemia, neuroferrittinopathy, Leigh’s disease, mitochondrial encephalopathy[6] . Hayflick et al studied 123 case and found PKAN2 mutation on patients with “eye of the tiger” sign.

MRI feature of our index case and his brothers were similar to radiological findings of Hallervorden-Spatz disease or PKAN. Genetic testing could not possible due to unavailability of test in our institution and differentiation from other Neurodegeneration associated with Brain Iron Accumulation (NIBA) could not be made.

There is no specific treatment for Hallervorder spatz disease. Management is only supportive and symptomatic. Drugs used for improvement of quality of life and dystonia are oral and intrathecal baclofen, trihexphenidyl, intramuscular botulinum toxin and oral deferiprone[7]. DBS is also considere in selected case of PKAN patient with drug resistant dystonia[8].

Conclusion

Hallervorder Spatz Disease or PKAN can be considered as a differential diagnosis when patient present with early onset behavior abnormality and movement disorder. The “eye of the tiger” pattern on MRI of brain is near specific for diagnosis of this disease and can identify presymptomatic siblings of affected patient.

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