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Radiologic Features of Hallervorden Spatz Disease

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Introduction

Hallervorden Spatz Disease is a rare familial neurodegenerative disorder, which primarily affects children but also can occur in adults¹. Major clinical features are abnormal involuntary movements and cognitive impairment². Diagnosis is based on clinical and radiological features. The radiological features are hypointense signals in globus pallidus and substantia nigra on MR! of brain³. Occasionally the pallidal hypointense signals surround hyperintense signals, this is known as “tiger-eye-sign”⁴ and is postulated to be specific for Hallervorden Spatz Disease. We report two brothers with such MRI findings.

Case Report

Case 1

A thirteen year, right handed boy presented to the neurology clinic with three year history of progressive walking difficulty and recurrent falls. He also suffered from abnormal involuntary movements, which would disappear during sleep and progressive dysarthria to the extent that his speech was barely intelligible. There was also a history of global cognitive decline. During the six month prior to presentation he became progressively irritable, restless and displayed sudden and inappropriate episodes of laughter. There was no history of fever, jaundice or seizures, His examination was significant for decreased comprehension and attention. His speech was hypophonic and barely intelligible. Generalized dystonia was noted, with ballistic component on right side in association with bilateral blepharospasm. His tone was increased bilaterally although more on the right side. Plantar response was normal. . His fundoscopic examination was normal. His CBC and liver function tests (LFTs) were normal, serum ceruloplasmin and urinary copper levels were also normal (0.243g/L and 23ug/24 hours respectively). He was given a trial of levodopa during 2nd year of his illness without any benefit. An MRI of the brain was performed, which revealed the classic “tiger-eye-sign” in the globus pallidus bilaterally (Figure 1).

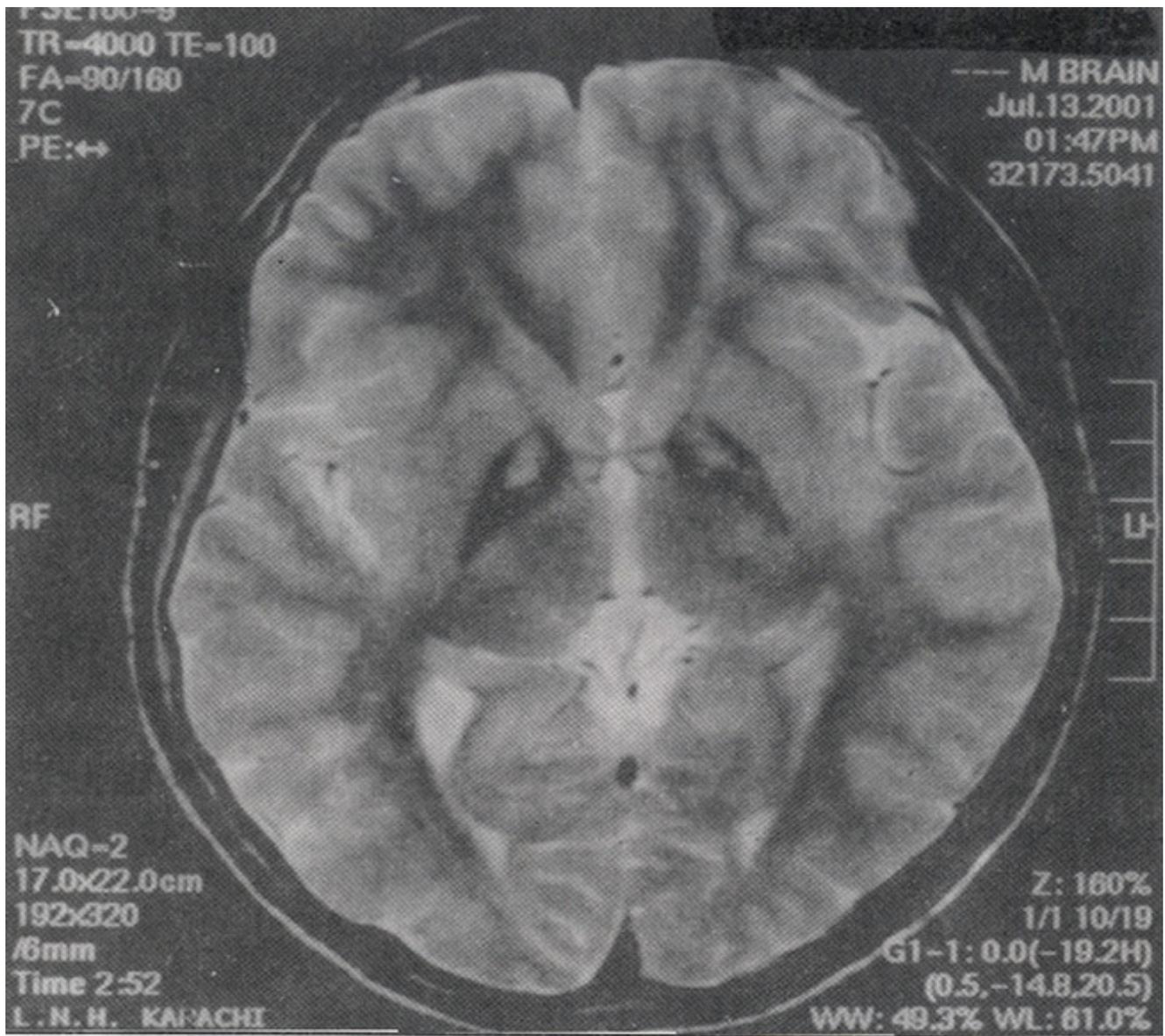


Figure 1. T2 (spin echo, TR 4000/TE 100) MRI of brain (patient 1) showing tiger-eye-sign.

Case 2

An eleven-year boy (younger brother of case 1) presented to neurology clinic with history of progressive walking difficulty, decreased school performance and involuntary movements for about two years. His examination was significant for mild cognitive impairment, generalized dystonia and mild irritability. His fundoscopic examination was also normal. His overall clinical presentation was similar to that of his elder brother (case 1) but was milder in severity. Laboratory workup including CBC, liver enzymes, serum ceruloplasmin and urinary copper levels were normal. His MRI brain showed similar findings i.e., tiger-eye-sign (Figure 2).

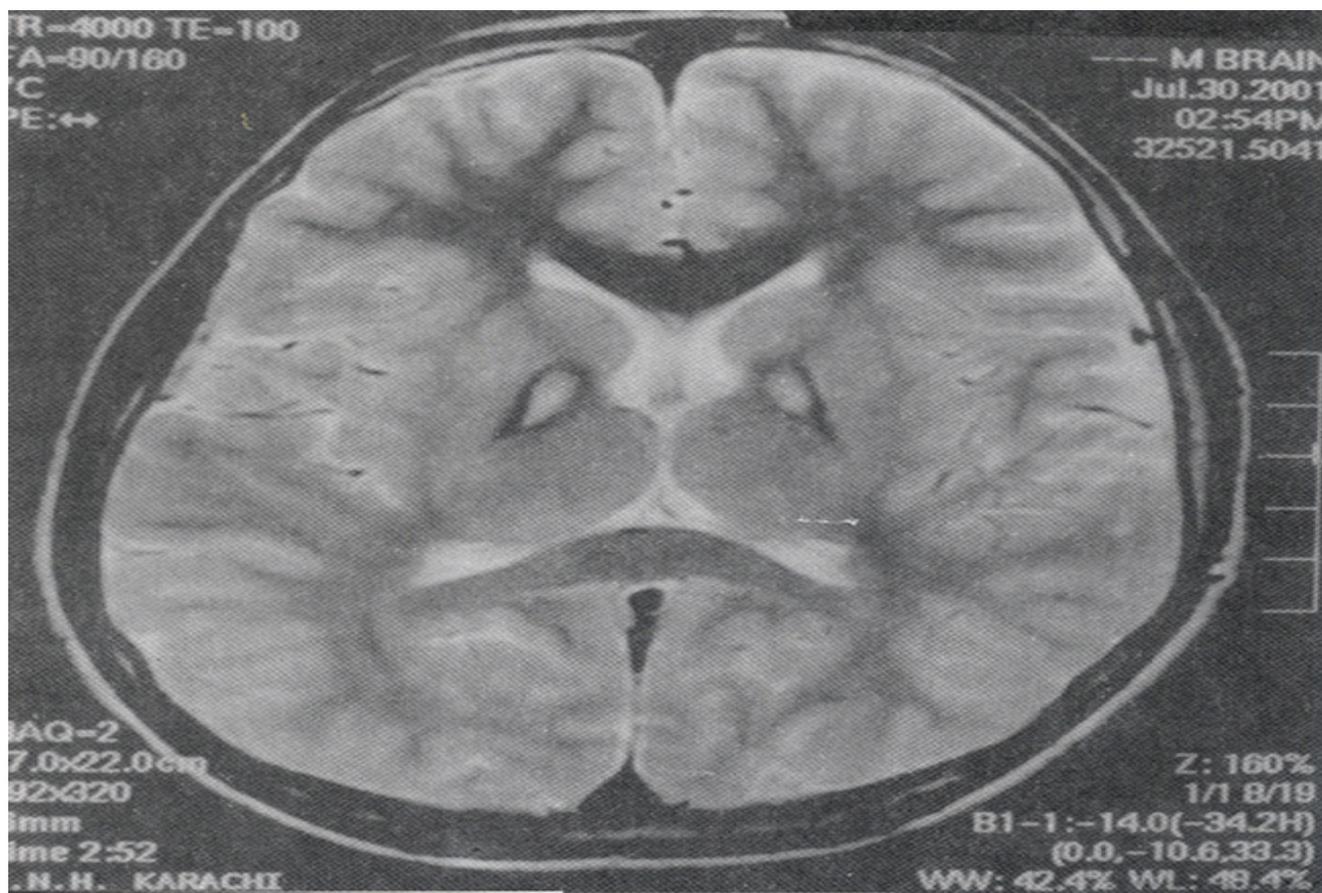


Figure 2. T2w (spin echo, TR 4000/TE 100) MRI of brain (patient 2) showing tiger-eye-sign.

Discussion

Hallervorden Spatz Disease is a rare autosomal recessive disorder, which was first described by Hallervorden and Spatz in a family in 1922¹. They described it as a familial disorder that began prior to age of 10 years, often associated with clubfoot deformity, gradually increasing stiffness in all limbs, impaired speech and dementia. Its gene is not identified yet though its location is chromosome 20⁵. The most important pathological findings are iron deposition symmetrically in both globi pallidi and substantia nigra (pars reticulata) and distal axonal swelling i.e. spheroids⁶. Clinical features include progressive gait differences, rigidity, spasticity, paucity of movement, dystonia, dysarthria, tremor and cognitive impairment². Retinitis pigmentosa is also present in 20% of patients³. Rarely blepharospasm, ballism, freezing, ptosis and apraxia of eyelid opening are seen³. The clinical presentation mimics several neurological disorders, the most important one being Wilson's disease. Differentiation between the two disorders is very important, as Wilson's disease is treatable whereas there is no treatment available for Hallervorden Spatz disease. The definitive diagnosis of Hallervorden Spatz disease could only be made on histopathological grounds hence the term Hallervorden Spatz syndrome has been used for the clinical entity, when diagnosis is made on the basis of clinical and laboratory parameters⁷. The most important investigation, which supports the diagnosis, is magnetic resonance imaging (MRI) of brain. Most common finding on MRI is symmetric hypointense signals in globus pallidus³. Sometimes hyperintense signals lie within these hypointense signals, the so-called "Tiger-eye sign"^{4,7,8}. This sign is said to be specific for the disease. Swieman devised the diagnostic criteria for the disease based on clinical and radiological and laboratory findings⁹. According to the

Swiaman criteria,

Hallervorden Spatz disease can be diagnosed if a person meets the obligatory criteria which are:

- a) onset during first two decades,
- b) progressive course and
- c) evidence of extra pyramidal dysfunction,

Along with the obligatory criteria, there must be present at least two of the following eight corroborative criteria:

- a) pyramidal tract signs,
- b) progressive mental retardation,
- c) seizures,
- d) retinitis pigmentosa/optic atrophy,
- e) positive family history,
- f) hypo densities in basal ganglia on MRI brain,
- g) abnormal cytoplasm in lymphocytes and
- h) sea-blue histocytes on bone marrow

We propose that application of these criteria may result in attributing false diagnosis of an untreatable and rare disease to the patients with treatable and comparatively common diseases like Wilson's disease and hence these criteria need to be revised. We suggest including the radiological findings as obligatory criterion. The diagnosis of Hallervorden Spatz Disease can be made confidently on the basis of the triad i.e. cognitive impairment, abnormal involuntary movements and the 'tiger-eye-sign' on MRI brain.

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