

# Hallervorden Spatz Syndrome: A Case Report

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## ABSTRACT

**Introduction:** Hallervorden-Spatz syndrome, now known as pantothenate kinase-associated neurodegeneration (PKAN), is an autosomal recessive disorder causing progressive extrapyramidal dysfunction and dementia. It is characterized by progressive degeneration of basal ganglia, globus pallidus and reticular part of the substantia nigra, produced by iron accumulation. The characteristic MRI brain pattern of HSD shows the “eye of the tiger” pattern.

**Case report:** Here we report 3 siblings where diagnosis was missed till MRI showed classic imaging findings. Till date, all patients with PKAN mutation whether classic or atypical had classic MRI findings suggesting thereby that MRI served as an important tool to predict mutation status. Treatment is symptomatic and requires combined effort of pediatrician, neurologist, ophthalmologist, physiotherapist, occupational therapist, geneticist and speech therapist.

**Conclusion:** This report is to sensitize clinicians regarding this entity and to differentiate it from other static and progressive neurological illnesses.

**Keywords:** Eye of tiger appearance, Dystonia, Neurodegeneration, PKAN

## INTRODUCTION

Hallervorden – Spatz disease is a rare neurological disorder which was earlier known as Neurodegeneration with brain iron accumulation.<sup>1</sup> It was 1<sup>st</sup> described by Hallervorden and Spatz in 1922<sup>2</sup> and characterised by familial brain degeneration with iron accumulation. A defect in pantothenate kinase 2 producing gene located in chromosome 20p13-p12.3 is reported in most of the cases, hence also termed as pantothenate kinase associated neurodegeneration (PKAN).<sup>3</sup> We report presence of this syndrome in 3 siblings (including two cousins) in a family with classic MRI findings.

## CASE REPORT

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 ap-

propriate 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 history of 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. Laboratory tests revealed no abnormality. MRI brain revealed area of hyperintensity within a region of hypointensity in medial globus pallidus bilaterally on T2 images -“eye of the tiger” pattern (Figure 1). 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. Serum electrolytes, iron, copper and ceruloplasmin levels were normal. Patient was given symptomatic treatment for spasticity with trihexphenidyl and baclofen, sodium valproate was given for seizures and clonazepam to reduce irritability. Physiotherapy treatment was also started. Seizures controlled and tone as well as spasticity decreased with significant improvement in sleep and oral intake after few weeks of treatment. On follow up the child is now accepting semisolids, no drooling with significant decrease in oromotor dyskinesia and generalized dystonias.

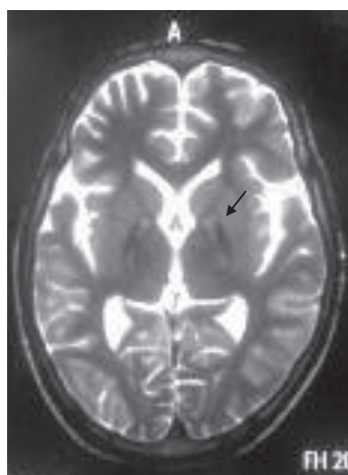
## DISCUSSION

Hallervorden – 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.<sup>4</sup> Dystonia, dysarthria, rigidity

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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 pallidus with a small area of central hyperintensity.(arrow)

and choreoathetosis is seen in 98% of cases, cortical tract signs in 25%, cognitive decline in 29%, optic atrophy in 3% and acanthocytosis in 3% cases. Hayflick et al<sup>1</sup> classified NBIA<sup>5,6</sup> in different groups on basis of age of onset and gene defect. The classical form, with PANK2 mutation, is characterized by early onset, rapid progression and presence of the typical eye-of-the-tiger sign. Our case had classic age of presentation and progression was also typical of the disease. Although seizures occur rarely in classic disease but were present in our case.

The eye-of-the-tiger sign on the MRI scan and clinical findings has contributed to a diagnosis of HSD for this patient. The characteristic MRI findings are symmetrical hyperintense signal changes in the anterior medial globus pallidus with surrounding hypointensity in the globus pallidus on bilateral sides on T2 – weighted images.<sup>7,8</sup> Management is usually symptomatic. Drugs used for spasticity and dystonia are baclofen and trihexphenidyl. Other therapies for relieving dystonia are intramuscular botulinum toxin, intrathecal baclofen, stereotactic pallidotomy,<sup>9</sup> bilateral thalamotomy and deep brain stimulation.<sup>10</sup>

## CONCLUSION

PKAN should be considered in differential diagnosis of early onset cognitive impairment and dystonia. The ‘eye of tiger’ sign is fairly specific and hence can be used to identify patients for PANK 2 genetic testing and has accurately identified presymptomatic siblings of affected children.

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