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EYE OF TIGER SIGN IN HALLERVORDEN SPATZ DISEASE (PANTOTHENATE KINASE II ASSOCIATED NEURODEGENERATION - PKAN): A RARE CASE REPORT.

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HOW TO CITE THIS ARTICLE:

Chethan BS, Yugandhara Shah. "Eye of tiger sign in Hallervorden Spatz disease (Pantothenate Kinase II associated neurodegeneration - PKAN): a rare case report". Journal of Evolution of Medical and Dental Sciences 2013; Vol2, Issue 50, December 16; Page: 9641-9644.

ABSTRACT:

INTRODUCTION: The term Hallervorden Spatz Disease is an autosomal recessive disorder. The patient presents with clinical features of progressive extra pyramidal dysfunction and dementia. The pathological triad observed in such disorders consists of iron deposition, axonal spheroids and gliosis in the globus pallidus. The eye of tiger sign allows the specific MR diagnosis of Hallervorden Spatz disease or related extrapyramidal parkinsonian disorders in the presence of supporting clinical signs. [4].

CASE PRESENTATION: A thirteen year old boy presented with progressive abnormal movements, intellectual decline, slowing of voluntary movement, dysarthria and occasional tremors. The patient had dystonia and rigidity on clinical examination. The patient was referred to Radiology department for MRI of the brain. The MR findings when correlated with clinical history helped us in making a diagnosis of Hallervorden Spatz disease, also known as Pantothenate Kinase 2 associated neurodegeneration (PKAN). The patient is now under conservative management.

CONCLUSION: The diagnosis of Hallervorden Spatz disease was made and the patient was further conservatively managed.

KEYWORDS: Pantothenate Kinase 2 associated neurodegeneration (PKAN), eye of tiger sign.

Clinical History: A thirteen year old boy presented with progressive abnormal movements, intellectual decline, and slowing of voluntary movement, dysarthria and occasional tremors. The patient had dystonia and rigidity on clinical examination.

Imaging findings: Axial T2 weighted image reveals marked hypointensity within bilateral globus pallidus with central hyperintensity. Axial T2 weighted FLAIR image also shows marked hypointensity within the globus pallidus bilaterally with central hyperintensity. Axial Diffusion weighted image also shows marked hypointensity in the globus pallidus with restricted diffusion centrally.

DISCUSSION: Hallervorden Spatz syndrome encompasses several disorders which share the clinical features of progressive extrapyramidal dysfunction and dementia with pathological triad of iron deposition, axonal spheroids and gliosis in the globus pallidum. Hallervorden Spatz disease is characterized by rigidity, dystonia, impaired postural reflexes and progressive dementia. Other distinct clinicopathologic disorders in the scope of Hallervorden-Spatz syndrome include dementia, tetraparesis, neurofibrillary tangles, retinitis pigmentosa and acanthocytosis with or without lipid abnormalities, including hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, pallidal

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degeneration [4]. Hallervorden Spatz disease also known as Pantothenate Kinase 2 associated neurodegeneration (PKAN), is a rare autosomal recessive condition [1].

The main pathology is excessive deposition of iron across the globus pallidus and substantia nigra. Iron deposition causes gliosis in the affected.

The main pathophysiologic basis of this disease is hypothetical. There is an enzymatic block in the metabolic pathway from cysteine to taurine.

The gene for the coding of pantothenate kinase is recently identified on chromosome 20p.12.3-13. This gene is required for the phosphorylation of pantothenic acid in the formation of coenzyme A. Defect in the phosphorylation leads to deficiency of Pantothenate kinase which in turn results in the underutilization of cystine.

The excess of cystine causes chelation of iron which leads to free toxic radical formation. The basal ganglia have excess of pantothenate receptors which explains the preferential affection of these areas resulting in excess iron deposition seen as diffuse hypointensity across these areas on the MR imaging. [2.]. The central hyperintensity is seen due to gliosis and spongiosis [2].

Clinically the patient may present at late childhood or early adolescence [5]. The most common presenting symptoms are gait or postural difficulties. The predominant neurologic features are extrapyramidal and includes dystonia, dysarthria, rigidity and choreoathetosis [6]. The course is progressive and most have a fatal outcome in 2 to 10 years [5].

On computed tomography, the findings are non specific. In some cases hyperdensity is noted across the globus pallidus.

MRI is the imaging modality of choice. The most characteristic finding on MR is the visualization of the "eye of tiger" sign on the T2 weighted / T2 weighted FLAIR images across the bilateral globus pallidus. The eye of tiger sign is due to marked hypointensity affecting the globus pallidus bilaterally and the central hyperintensity is due to gliosis and spongiosis [2].

Diffusion imaging seems to show nonspecific changes with low signal intensity, due to susceptibility effects. ADC values were slightly increased compared to normal appearing thalamus [3].

Treatment of Hallervorden Spatz disease needs a multidisciplinary approach. A team of neurologist, ophthalmologist, physiotherapist, occupational therapist and speech therapist is involved. The treatment is symptomatic. Baclofen and trihexyphenidyl are most effective drugs for disabling dystonia and spasticity. Regular physiotherapy is important for normalization of muscle tone and improving functional independence. Passive stretching, reflex inhibitory postures, weight bearing and sensory stimulation with many more approaches help these patients to remain functional skills and improve quality of life [5].

Final diagnosis: Pantothenate Kinase 2 associated neurodegeneration (PKAN), also known as Hallervorden Spatz disease.

Differential diagnosis:

extrapyramidal parkinsonian disorders
aceruloplasminemia and neuroferritinopathy

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Fig. 1: Axial T2 weighted image reveals marked hypointensity within bilateral globus pallidus with central hyperintensity.

Fig. 2: Axial T2 weighted FLAIR image also shows marked hypointensity within the globus pallidus bilaterally with central hyperintensity.

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Fig. 3: Axial Diffusion weighted image also shows marked hypointensity in the globus pallidus due to susceptibility artifacts with restricted diffusion centrally.

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Date of Submission: 16/11/2013.
Date of Peer Review: 18/11/2013.
Date of Acceptance: 23/11/2013.
Date of Publishing: 10/12/2013