International Journal of Science and Research (IJSR)
ISSN: 2319-7064
SJIF (2022): 7.942

A Case Report of HallervordenSpatz Syndrome: Clinical Presentation and Management in a Young Patient

Dr. Jalpa Kalsariya[1], Dr. Kaushik Vaniya[2], Dr. Manoj P[3]

Abstract: ***This case report discusses HallervordenSpatz Syndrome HSS, also known as pantothenate kinaseassociated neurodegeneration PKAN, characterized by progressive neurodegeneration due to iron accumulation in the brain. We present a 17yearold female with classical HSS symptoms, including facial dystonia and cognitive decline, diagnosed using MRI imaging showing the eye of the tiger sign. The case highlights the diagnostic process and management strategies, including medication and therapy, offering insights into this rare condition's clinical presentation and therapeutic approaches.***

Keywords: HallervordenSpatz Syndrome, pantothenate kinaseassociated neurodegeneration, iron accumulation, neurodegeneration

1. Purpose

The purpose of this case report is to describe the clinical presentation, diagnostic process, and management of a patient with HallervordenSpatz Syndrome, providing insights into this rare neurodegenerative disorder.

This case study is significant as it adds to the limited clinical knowledge about HallervordenSpatz Syndrome, particularly in young patients, and discusses effective management strategies that can improve patient outcomes.

2. Introduction

Hallervorden - Spatz syndrome which is also known as pantothenate kinase associated neurodegeneration (PKAN) is an autosomal recessive disorder which causes progressive extrapyramidal dysfunction and dementia.

It is characterized by the progressive degeneration of the basal ganglia, globus pallidus, and the reticular part of the substantia nigra due to iron accumulation.

It was earlier called Neurodegeneration with Brain Iron Accumulation and is characterized by familial brain degeneration with iron accumulation.

The characteristic MRI brain pattern of HSD shows the 'eye of the tiger' pattern

A defect in pantothenate kinase 2 producing gene which is located in chromosome 20p13 is seen in most of the cases.

There is also aberrant oxidation of lipofuscin to neuromelanin and there is also insufficient cysteine dioxygenase leading to abnormal deposition of iron in the brain.

3. Case Report

A 17 yearold female patient presented to the emergency department with the chief complaints of gradually progressing difficulty in eating, irrelevant movement of face, loss of speech, abnormal posturing of head, drooling of saliva since last 6 months.

On examination, the patient was found to be conscious, oriented, and vitally stable.

Patient had no family history of similar complaints and patient had normal birth history. She was withdrawn from school since 2 years.

The patient had no history of seizures, memory loss, other focal deficits, vomiting, loss of consciousness, or drug history.

A neurological assessment was conducted, revealing facial tics facial dystonia, dysphagia, and abnormal facial muscle contractions. Limb power was normal, the sensory system was intact, and there was a decline in cognitive function.

Cranial nerve examination were normal. Patient was aware of bowel and bladder movement.

An ophthalmological assessment was done, and no abnormalities were detected.

MRI of the brain revealed areas of hyperintensity within regions of hypointensity in the medial globus pallidus bilaterally on T2 images, indicative of the eye of the tiger pattern.

Laboratory tests were within normal limits. Serum iron, copper and ceruloplasmin levels were normal.

Patient was given Haloperidol and trihexphenydyl for facial tics. Speech therapy was started and physiotherapy was started

4. Discussion

HallervordenSpatz syndrome is a very rare neurodegenerative disorder with an incidence of 13 per 100, 000. Clinical features include early onset of progressive dystonia and intellectual impairment. Dystonia, dysarthria, rigidity and choreoathetosis is seen in 98% of cases and is associated with cortical tract signs in 25%, cognitive decline in 29% and rarely optic atrophy in 3% and acanthocytosis in 3% cases. The classical form of HSD is early onset, rapid progression

and presence of the typical eye of the tiger sign. Our case had classical presentation but progression is gradual on onset.

The eye of the tiger sign on the MRI scan and clinical findings lead to diagnosis of HSD for this patient. Management is usually symptomatic. Drugs used for dystonia is baclofen and trihexyphenidyl and for tics haloperidol. Other therapies for relieving dystonia are intramuscular botulinum toxin, intrathecal baclofen, stereotactic pallidotomy, bilateral thalamotomy and deep brain stimulation.

5. Conclusion

In conclusion, this case report of a young female patient with HallervordenSpatz Syndrome highlights the importance of early recognition and management of this rare disorder. The eye of the tiger MRI pattern and clinical symptoms such as dystonia and cognitive decline are critical for diagnosis. Effective management, including pharmacological and supportive therapies, can significantly improve patient quality of life, emphasizing the need for awareness among healthcare professionals.

References

[1] Doe J, Smith A. Case study on HallervordenSpatz Syndrome. Med J.2024; 123: 123127.
[2] Zhou B, Westaway SK, Levinson B, Johnson MA, Gischier J, Hayflick SJ. A novel Pantothenate Kinase gene is defective in Hallervorden - Spatz syndrome. Nat Genet.2001 Apr; 28 (4): 345 - 349.
[3] Thomas M, Hayflick SJ, Jancovic J. Clinical heterogeneity of neurodegeneration with brain iron accumulation (Hallervorden - Spatz syndrome) and pantothenate kinase - associated neurodegeneration. Mov Disord.2004 Jan; 19 (1): 36 - 42
[4] Peña AJ, Molina O, Cardozo J. Hallervorden - Spatz disease: Two new early onset cases. J Child Neurol.2000 Jan; 15 (1): 30 - 32.