Case Report

HALLERVORDEN-SPATZ DISEASE - A RARE CASE REPORT - "Eye of the Tiger" Sign

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ABSTRACT

Background: Hallervorden-Spatz disease (HSD) is a rare neurological disease characterized by progressive degeneration of basal ganglia, globuspallidus and reticular part of the substantianigra, produced by iron accumulation. The defect has been found in the pantothenate kinase 2 (PANK2) producing gene located in chromosome 20p13-p12.3. Clinical presentations include dystonia, dysarthria, dysphasia, dementia, severe mental retardation and severe movement disability may develop at later stages. Rare clinical features include rigidity, choreoathetosis, seizures, optic atrophy and pigmentary retinopathy. The characteristic MRI brain pattern of HSD shows the "eye of the tiger" pattern. Treatment is symptomatic. We present the case of a patient, 19 years old boy with Hallervorden-Spatz disease who came to our physiotherapy department with features of spasticity, dystonia and gait difficulty. He was diagnosed on the basis of clinical findings and typical MRI brain of "eye of the tiger" pattern. His detailed evaluation was carried out and physiotherapy treatment was started.

KEY WORDS: Hallervorden-Spatz disease; eye of the tiger; iron accumulation.

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Access this Article online

Quick Response code

International Journal of Physiotherapy and Research
ISSN 2321- 1822
www.ijmhr.org/ijpr.html

Published: 11 August 2013

Received: 01 July 2013

Accepted: 16 July 2013

INTRODUCTION

Hallervorden-Spatz disease (HSD) is a rare neurological disease described in 1922 by Julius Hallervorden and Hugo Spatz characterized by progressive degeneration of basal ganglia, globus pallidus and reticular part of the substantia nigra, produced by iron accumulation, with normal levels of iron in blood and cerebrospinal fluid. Also termed as neurodegeneration with brain iron accumulation (NBIA).[1] In most of the cases, a defect has been found in the pantothenate kinase 2 (PANK2) producing gene located in chromosome 20p13-p12.3 and hence this disease is also termed as pantothenate kinase associated neurodegeneration (PKAN).

The onset of symptoms most commonly occurs in late childhood or early adolescence between ages 7 and 15 years. It is inherited as an autosomal recessive genetic trait, although 15% of the patients are sporadic. It occurs in all races and has a similar frequency in both genders. The course is progressive and most have a fatal outcome in 2 to 10 years.[2] Pathological findings in brain include rust brown pigmentation due to iron deposition, axonal swellings or spheroids and neuronal loss with gliosis predominantly in the globus pallidus and pars reticularis of the substantia nigra.[3]

The exact etiology of HSD is not known. One proposed hypothesis is that abnormal peroxidation of lipofuscin to neuromelanin and deficient cysteine deoxygenase lead to abnormal

iron accumulation in the brain. While portions of the globus pallidus and pars reticulata of substantia nigra (SN) have high iron content in healthy individuals, individuals with HSD have excess amounts of iron deposited in these areas.[4]

Clinical presentations include dystonia, dysarthria, dysphasia, dementia, severe mental retardation and severe movement disability especially gait difficulty may develop at later stages. Rare clinical features include rigidity, choreoathetosis, seizures, optic atrophy and pigmentary retinopathy.[5] Radionuclide scan reveals increased iron uptake in the basal ganglia. The characteristic MRI brain pattern of HSD showing area of hyperintensity within a region of hypointensity in the medial globuspallidus bilaterally on T_2 weighted images [the "eye of the tiger" pattern] corresponds to the pathological findings. The hypointensity on T2 weighted image is because of iron deposition and central hyperintensity is secondary to gliosis and spongiosis.[6]

The treatment of patients with Hallervorden-Spatz disease (HSD) remains mostly symptomatic. A multidisciplinary team approach involving physical, occupational and speech therapists may be needed to improve functional skills and quality of life.

CASE REPORT

A 19 years old boy came to our Physiotherapy department with abnormal posturing of head and limbs and difficulty in walking since 5 and half years. He had history of stiffness in lower limbs progressing to the upper limbs. There was progressive difficulty in walking with frequent falls for the past 2 years but he could still ambulate with assistance. There was a history of seizures since last 3 years and had a progressive decline in scholastic performance.

His pre and post birth history was uneventful and there was no delay in attaining the milestones. Family history was not significant. On Observation, posture was dystonic with all limbs flexed. On physical examination he was alert and cooperative. No abnormality of the cranial nerves was found. Speech disturbances including dysarthria and drooling was also present. There was a generalised increase in tone of all four limbs with spasticity more apparent in lower limbs. Tendoachilles, hamstrings and adductors had grade 2 spasticity on Modified Ashworth scale. Muscle strength was above grade 3 for all muscles on Manual muscle testing. Deep tendon reflexes showed mild hyperreflexia with bilateral extensor plantar response. Gait examination revealed problems with both stance and swing pattern of gait and requires assistance to walk. Coordination was normal bilaterally. Sensory system revealed no abnormality.

Laboratory tests revealed no abnormality. Slit lamp examination of the eye did not reveal Kayser-Fleischer ring. MRI of the brain revealed area of hyperintensity within a region of hypointensity in the medial globuspallidus bilaterally on T_2 weighted images - the "eye of the tiger" pattern. The patient was given symptomatic treatment with anticholinergics and muscle relaxants (baclofen) and sodium valproate was continued.

DISCUSSION

Hallervorden-spatz disease is a rare disorder characterized by progressive extrapyramidal dysfunction and dementia. The eye-of-the-tiger sign on the MRI scan and clinical findings has contributed to a diagnosis of HSD for this patient.

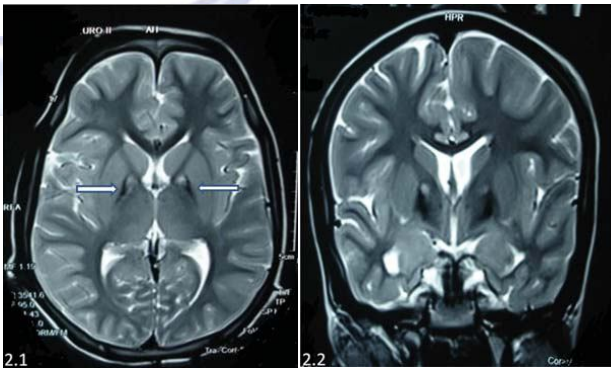

Figure 1: MRI brain showing T2 axial (2.1) and coronal (2.2) images: Bilateral symmetrical hypointensity in globus pallidus with central hyperintensity "eye of tiger sign''.

Treatment requires multidisciplinary approach including neurologist, ophthalmologist, physiotherapist, occupational therapist, geneticist and speech therapist. Treatment of Hallervorden-Spatz disease (HSD) is symptomatic. Baclofen and trihexyphenidyl remain the most effective drugs for disabling dystonia and spasticity. Baclofen is a derivative

of gamma-aminobutyric acid (GABA). It is primarily used to decrease spasticity. Botulinum toxin can be helpful for many patients, especially those whose quality of life is improved by treating a limited body region. For example, injections in the facial or orobucculingual muscles can greatly improve speech and eating abilities. When oral baclofen is no longer able to adequately control the movement disorder, placement of a continuous intrathecal baclofen pump may be considered.

Deep brain stimulation (DBS) is also an option for relieving some symptoms. A recent report by Castelnau et al on the use of DBS in six PKAN patients showed overall improvements in writing, speech, walking, and global measures of motor skills.[7] If there is difficulty in swallowing and nutrition is impaired, gastrostomy placement may prove necessary. Oral glycopyrrolate and hyoscine patch may be useful for drooling. As PKAN is autosomal recessive in inheritance, at each conception sibling has 25% chance of being affected. Prenatal diagnosis may be possible if facilities of genetic testing are available.

Regular physiotherapy for normalization of muscle tone and improving functional independence is necessary. Passive stretching, reflex inhibiting postures, weight bearing, sensory stimulation, physio ball therapy and many more approaches help such patients to regain some functional skills and improve quality of life.

CONCLUSION

Dystonic posturing is the most prominent feature in Hallervorden-Spatz disease (HSD), it can be reduced through EMG Biofeedback, sensory stimulation, weight bearing and approximation, biomechanical loading by distal fixation with weights, relaxation techniques. Spasticity and rigidity can be reduced by prolonged positioning, splinting or casting, functional electrical stimulation, biofeedback and proprioceptive neuromuscular facilitation.

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How to cite this article:
Mallikarjunaiah H.S, Kanchan Agrawal, Pravin Aaron. Hallervorden-spatz disease - A rare case report. Int J Physioth Res 2013;03:107-09.