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Case Report

Pantothenate kinase associated neurodegeneration in late adulthood: an atypical case from India

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ABSTRACT

Pantothenate kinase associated neurodegeneration (PKAN) is a rare autosomal recessive disorder and the most common subtype of neurodegeneration with brain iron accumulation (NBIA). While classical PKAN typically presents in childhood, atypical forms may manifest later in life with heterogeneous neurological and psychiatric features, often resulting in diagnostic delay. We report a case of atypical PKAN in a 45-year-old woman who presented with a three-year history of progressive dysarthria, involuntary tongue protrusion, sialorrhea, difficulty in jaw closure, and involuntary movements of the right upper limb. Neurological examination revealed spastic dysarthria, oromandibular dystonia, orolingual chorea, rigidity, resting tremor, hyperreflexia, and a positive Hoffmann sign. Psychiatric manifestations included anxiety, depressive episodes, and impulsivity. Routine laboratory investigations were unremarkable except for mild iron deficiency anaemia. Magnetic resonance imaging of the brain demonstrated the characteristic bilateral "eye of the tiger" sign in the globus pallidus, strongly supporting the diagnosis of PKAN. The patient was treated with trihexyphenidyl, tetrabenazine, botulinum toxin injections, and supportive therapy. This case highlights the importance of considering PKAN in the differential diagnosis of adult onset movement disorders with psychiatric manifestations. Recognition of its characteristic neuroimaging features is essential for timely diagnosis, appropriate symptomatic management, and genetic counselling. The rarity of adult onset PKAN and its diverse clinical presentation underscore the need for greater awareness among clinicians.

Keywords: Pantothenate kinase associated neurodegeneration, Neurodegeneration with brain iron accumulation, Eye of the Tiger sign, Atypical PKAN

INTRODUCTION

Pantothenate kinase associated neurodegeneration (PKAN) is an autosomal recessive disorder that falls under the broad spectrum of conditions known as neurodegeneration with brain iron accumulation (NBIA). It is also the most prevalent form of NBIA, accounting for nearly half of all NBIA diagnoses.[1-3] While exact prevalence data are lacking, the condition is considered extremely rare, with estimates ranging from 1–3 per 1,000,000 populations.[4-6]

The disease is caused by mutations in the pantothenate kinase 2 (PANK2) gene, located on chromosome band 20p13.[7,8] It was earlier called the Hallervorden Spatz disease and is characterized by the pathological deposition of iron in the globus pallidus.[3,5,6]

The PANK2 gene encodes pantothenate kinase, an enzyme that role in iron metabolism by maintaining redox reactions. Mutations in this gene can lead to pathological iron accumulation and free radical induced brain damage.[8,9]

Our patient presented to the medicine outpatient department with a three-year history of dysarthria and involuntary movements in the right upper limb. Subsequent investigations revealed the characteristic 'eye

of the tiger' sign on magnetic resonance imaging (MRI). Only a limited number of cases have been reported in the literature worldwide, with even fewer documented from India emphasizing the rarity of the condition and the uniqueness of this case report.

This case report aims to provide a comprehensive analysis of a patient with PKAN, highlighting the diagnostic challenges, clinical manifestations, and available treatment options.

CASE REPORT

A 45-year-old female presented to medicine outpatient department of a tertiary care centre in western India, with history of progressive worsening of symptoms including difficulty in speaking (dysarthria), involuntary protrusion of tongue, excessive drooling of saliva, difficulty in closing jaw and involuntary movements of the arm and forearm on the right side. On examination involuntary muscular contractions of the tongue and mandibular muscles were noted, indicating oromandibular dystonia. She also reported worsening of difficulty in chewing. There was no significant family history for similar neurological complaints and no history of focal neurological deficit, bowel and bladder disturbances and head trauma.

The past medical history was significant for stuttering since childhood. Additionally, she also complained of mild depressive episodes, anxiety, and impulsivity.

Examination and investigation

On examination, the patient was well oriented to time, place, and person. Patient was conscious, cooperative and obeying commands. Her vital signs were normal. Physical examination revealed mild pallor and koilonychia. She had dysarthric (spastic) speech. Mini mental state examination (MMSE) score was 25/30 (normal). Cranial nerve examination was normal. Motor examination revealed rigidity in right upper arm. Deep tendon reflexes were exaggerated (biceps, triceps, supinators, ankle, knee). Hoffmann sign was elicited and was found positive.

The right upper limb exhibited resting tremors characterized by a regular rhythm and coarse amplitude. Additionally, orolingual chorea was observed, presenting with an irregular rhythm and coarse amplitude.

Haematological investigations indicated mild iron deficiency anaemia, supported by the presence of microcytic, hypochromic red blood cells on peripheral smear examination.

T2 weighted MRI of the brain demonstrated bilateral hyperintense areas surrounded by hypointense zones in the globus pallidus, consistent with the characteristic 'eye of the tiger' sign (Figure 1).

Treatment

The patient was treated with oral trihexyphenidyl, starting at a low dose and gradually increased by 2 mg every three days. Oral tetrabenazine was also prescribed at 25 mg per day for the first five days and then gradually increased. Additionally, botulinum toxin type A was administered—50 units into the genioglossus muscle and 25 units into each of the bilateral lateral pterygoid muscles. The treatment plan also included multivitamin B complex, folic acid, and iron supplements. Currently, the patient is on regular monthly follow up.

Table 1: Laboratory investigations.

Haematological investigations	Result/findings
CBC	Mild iron deficiency anaemia
Peripheral smear	No acanthocytes seen microcytic hypochromic RBCs are seen
RFT	Within normal limits
LFT	Within normal limits
Electrolytes	Within normal limits
Thyroid profile	Within normal limits
RBS	124 mg/dl
S. Ceruloplasmin	37.75 mg/dl (20-40 mg/dl)
S. Ferritin	22 ng/ml (13-150 ng/ml)

CBC-Complete blood count; RBC-red blood cells; RFT-renal functions test; LFT-liver functions test; RBS-random blood sugar; mg-milligram; dl-decilitre; ng-nanogram; ml-millilitre

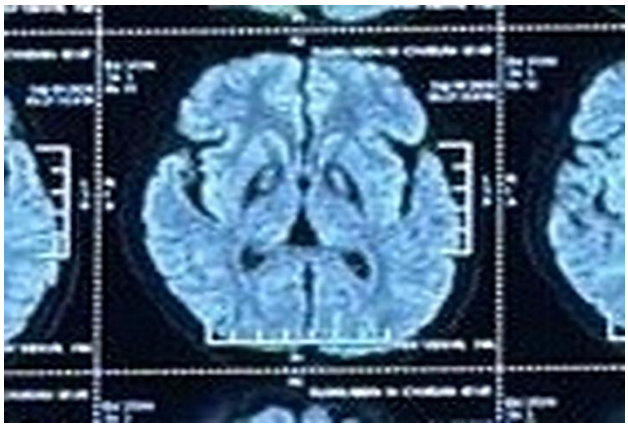

Figure 1: "Eye of the tiger sign" seen in MRI.

DISCUSSION

PKAN as a disease has a wide spectrum of presentations, including motor abnormalities (pyramidal and extrapyramidal features), intellectual impairment, psychiatric features, along with overlapping features of parkinsonism, leading to misdiagnosis. On the basis of presenting features, it can be classified into typical and atypical PKAN.

The majority of case reports described in the literature are of typical PKAN, which has onset in first decade and has early gait impairment, pyramidal features, extrapyramidal features (limb dystonia and oromandibular dystonia), no psychiatric symptoms or cognitive decline. Fundoscopy in 2/3rd of cases reveals pigmentary retinopathy. The lifespan of the child is significantly reduced.

The presence of orolingual dystonia, leading to dysphagia, dysarthria, palilalia, involuntary tongue protrusions and sialorrhea can be seen in both cases but is classically associated with atypical PKAN. Additionally, they also have psychiatric symptoms, cognitive decline and later age of onset. They have prolonged duration of symptoms, late gait impairment and no fundoscopic findings.

The peripheral smear may reveal acanthocytes in any of them. The radiological finding namely the characteristic “eye of the tiger” sign is seen in both.

Although genetic confirmation of a PANK2 mutation would have strengthened the diagnosis, genetic testing could not be performed because of resource constrained settings. The diagnosis was therefore based on the characteristic clinical and radiological findings.

In our case, the female had prolonged symptoms including stuttering from childhood, presenting complaints like oromandibular dystonia, orolingual chorea, spastic dysarthria, pyramidal features (brisk reflexes, bilateral extensor plantar, positive Hoffman sign), extra pyramidal features (chorea, dystonia), features of Parkinsonism (rigidity, bradykinesia and resting tremors in right upper limb) and psychiatric ailments along with the classical radiological sign lead to the diagnosis.

CONCLUSION

This case underscores the importance of considering PKAN in the differential diagnosis of adult onset movement disorders, even in patients presenting beyond the third decade of life. Although PKAN is typically inherited in an autosomal recessive manner and often presents in childhood, atypical forms can manifest later in adulthood with a broad spectrum of neurological and psychiatric features. In this patient, the absence of a positive family history did not preclude the diagnosis. The constellation of clinical findings, including facial and orolingual dystonia, parkinsonism, and significant psychiatric symptoms raised clinical suspicion. The diagnosis was further supported by characteristic MRI findings, including the pathognomonic "eye of the tiger" sign on T2 weighted imaging.

This case highlights the necessity for comprehensive clinical and neuroimaging evaluation in patients with complex extrapyramidal syndromes and psychiatric manifestations. Given its rarity and variable presentation, PKAN is often underdiagnosed or misdiagnosed and should be included in the differential diagnosis of overlapping movement and psychiatric disorders. Diagnostic delays are common, and the actual number of patients may be higher than reported.

The early diagnosis of atypical PKAN enables timely initiation of physiotherapy and prompt, symptom directed therapy, such as the use of anticholinergics, benzodiazepines, and baclofen for managing parkinsonism, spasticity, and movement disorders. Botulinum toxin may also be used for focal dystonia. Furthermore, it facilitates genetic counselling and the implementation of a coordinated, multidisciplinary management plan.

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