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Case Report

A Rare Case of Progressive Ataxia with Palatal Tremors

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Abstract

Palatal tremors are subdivided into essential (EPT) and symptomatic (SPT) forms according to clinical and etiological features. A subgroup of the SPT form has a syndrome of progressive ataxia and palatal tremor (PAPT). PAPT may be divided into sporadic and familial forms. In sporadic PAPT patients typically have olivary hypertrophy and increases signal intensity on T2-weighted MRI and progressive cerebellar degeneration. Familial form shows marked atrophy of cervical cord and brainstem with corticospinal signs, the hypertrophic olivary appearance on MRI will be absent. Few clinical features and MRI characteristics can help in differentiating familial and sporadic form even when genetic tests are inconclusive.

Here we describe a rare case of Progressive Ataxia with Palatal Tremors(PAPT) with features suggestive of familial form with inconclusive genetic tests.

Keywords: Palatal Tremors; progressive ataxia; olivary hypertrophy

Introduction

Progressive ataxia with palatal tremors is a rare neurodegenerative disease that is classified into familial and sporadic forms. The main clinical feature is palatal tremors, formerly called as palatal myoclonus. It is subdivided into essential (EPT) and symptomatic (SPT) forms. Symptomatic palatal tremors are associated with lesions in Guillain-Mollaret triangle.

Here we present a case of 43year old male presented with progressive ataxia, dysphagia, dysarthria with palatal tremors and with features suggestive of familial form of PAPT.

Case Report

Patient information

A 43-year-old man with no comorbidities, presented with 3-years history of unsteadiness of gait which was gradually progressive and 2-years history of nasal regurgitation of foods, dysarthria and 3-months history of involuntary rhythmic movements of the palate and head nodding. There was no pertinent past medical history. He also had family history of tremors and unsteadiness to his maternal grandfather which was not evaluated and he expired at his 60's due to myocardial infarction.

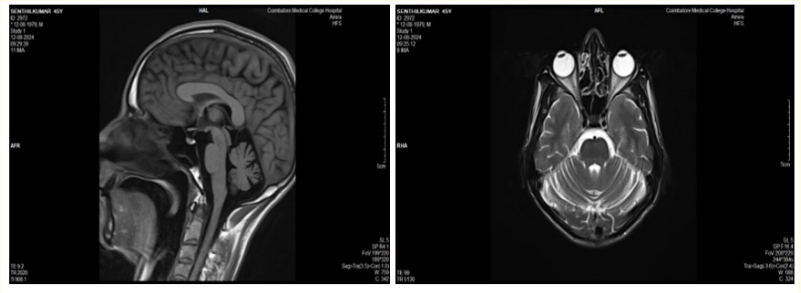
Clinical findings

Patient was conscious, oriented with scanning speech and bilateral symmetrical palatal tremor of frequency 140- 160/min (Video 1). Uvula in midline, gag reflex normal. On spino-motor examination spasticity of all 4 limbs and brisk deep tendon reflexes were seen with normal power. Wide based ataxic gait was present with no dysmetria. Rest of the clinical examination revealed no abnormalities.

Diagnostic assesment

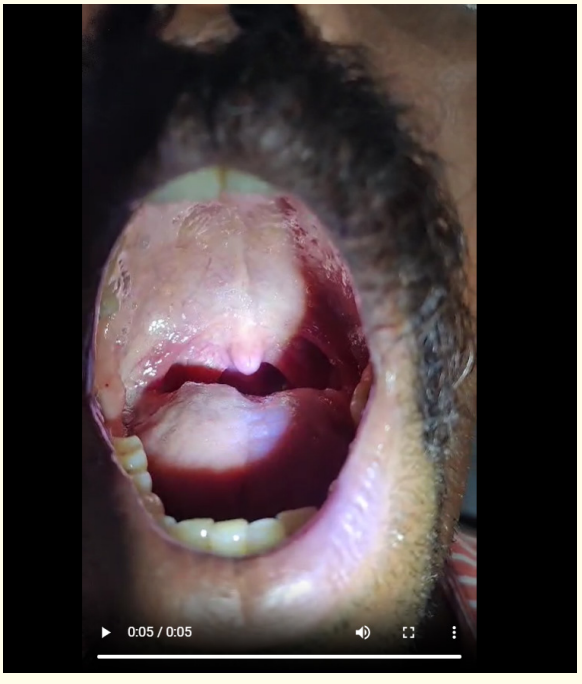
Investigations revealed normal routine blood analysis. TSH, antinuclear antibodies, CT thorax and abdomen, 2D-ECHO were normal. Stool examination, UGI endoscopy to rule out celiac disease was done which were normal. CSF examination and NCS was normal. MRI brain revealed mild atrophic changes in brainstem and cerebellum (Figure 1A). No olivary hypertrophy (Figure 1B). Mild thinning of bilateral corticospinal tracts was noted in MR tractography.

Whole genome sequencing was negative. Heterozygous for uncertain significance variant in the ITPRI gene and CCDC88C gene associated with SCA 15 and 40 was seen (Figure 2). However it doesn't test for some of the relevant genes like POLG.



A) B)

Figure 1: A) MRI brainT1W sagittal section showing mild brainstem and cerebellar atrophy
B) MRI T2W axial Image at medullary level which is normal.



Video 1

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Name:	[redacted]	Case ID:	40900112644
Age:	45 years	Sample Type:	Peripheral Venous Blood
Sex:	Male	Sample Collection Date:	23 Apr 2024
Referring Clinician:	[redacted]	Sample Receipt Date:	24 Sep 2024
Test Required:	Whole Genome sequencing	Report Date:	23 Oct 2024
Location:	[redacted]	Version:	1

CLINICAL INFORMATION/HISTORY

[redacted] presented with insidious, slowly progressive ataxia, palatal tremor and dysphagia. There is a previous history of injury to the right hand and finger seven years back for which a surgery was done and a family history of the patient's maternal grandfather also having a history of unsteadiness. On examination, he was found to have swaying on both the sides when walking, involuntary movements of jaw and throat, slurring of speech, nasal regurgitation, head nodding, spasticity of all limbs, phonatory gap, watering of right eye , conjunctivitis for a week, impaired tandem gait and impaired heel shin test. USG of the abdomen revealed hemangioma and MRI Brain revealed T2 Flair hyperintensity in the anterior region of the medulla with diffusion restriction, mild cerebellar and brainstem atrophy, subacute infarct and demyelination. Tractography showed mild thinning of bilateral corticospinal tract initially there were no gait issues but later he was unable to do work due to unsteadiness. There is no history of weakness of the limbs. He has been evaluated for the gene variations related to the reported phenotype.

The following HPO terms were used for analysis: progressive gait ataxia, palatal tremor, dysphagia, involuntary movements, slurred speech, nasal regurgitation, jerky head movements, spasticity, conjunctivitis, impaired tandem gait, impaired heel walking ability, hemangioma, cerebellar atrophy, atrophy/degeneration affecting the brainstem, abnormal corticospinal tract morphology

RESULT SUMMARY

No pathogenic or likely pathogenic variants causative of the reported phenotype were identified.
*Correlation with clinical profile and family history is required.

Summary of Findings

Variants Potentially Relevant to the Indication for Testing:

The index patient is:

- Heterozygous for a Uncertain Significance variant in the *ITPR1* gene associated with SPINOCEREBELLAR ATAXIA 15; SCA15 .
- Heterozygous for a Uncertain Significance variant in the *CCDC88C* gene associated with SPINOCEREBELLAR ATAXIA 40; SCA40 .

Carrier Status:

No Pathogenic or Likely Pathogenic variants were detected in the Carrier gene list.

Secondary Findings (AGMG gene list):

No Pathogenic or Likely Pathogenic (Class 1/2) variants were detected in the ACMG gene list.

Figure 2: Genetic test report.

Intervention and follow up

Patient was treated with clonazepam, carbamazepine, trihexyphenidyl for palatal tremors. After a few days frequency of palatal tremors decreased from 140 – 160 range to 90 to 110 range. However there is no improvement in ataxia.

Discussion

Palatal tremor is an involuntary, rhythmic and oscillatory movement of the soft palate. It can be unilateral or bilateral. Palatal tremors can have huge variability of frequency (20-420 cycles/in) [1]. It is subdivided into essential (EPT) and symptomatic (SPT) forms according to clinical and etiological features [2]. EPT occurs without an obvious structural lesion in the brain [3] and is characterized by rhythmic movement of anterior soft palate, attributable to contractions of the trigeminal nerve–innervated tensor veli palatini muscle, and not uncommonly auditory clicks[2]. SPT is often associated with a brainstem or cerebellar lesion involving the dentato-rubro-olivary pathway (Guillain-Mollaret triangle) [4]. In SPT, palatal movements result from activation of the levator veli palatini muscle.

GMT has three vertices: Red nucleus(RN) within the midbrain, inferior olivary nucleus (ION) in the medulla, and dentatae nucleus (DN) in the contralateral cerebellum. It has three interconnecting edges: descending "rubro-olivary fibers" along the central tegmental tractus from the RN to the ipsilateral ION, crossing "olivocerebellar fibers" between the ION and the contralateral cerebellar cortex via the inferior cerebellar peduncle, and "dentatorubral fibers" ascending from the DN to the contralateral RN via the contralateral superior cerebellar peduncle to the original RN to complete the DRO by decussating in the midbrain [5]. Any injury involving one of the vertices or edges of the triangle results in degeneration, which initially causes hypertrophy of the ION followed by its atrophy, called hypertrophic olivary degeneration, which typically presents with clinical symptoms of cerebellar dysfunction, dysmet-

ria, palatal tremor, or ataxia [3]. The cerebellum is predominantly excitatory, but the dentate nucleus within it has the primary function of inhibiting the inferior olivary nucleus via GABAergic projections. If the triangle is interrupted by pathology (vascular insult, demyelination or tumour), the inferior olivary nucleus is no longer inhibited and it hypertrophies [6]. As the inhibition is lost olivary nucleus rhythmically discharges causing palatal tremors.

A subgroup of the SPT form has a syndrome of progressive ataxia and palatal tremor (PAPT) [2]. PAPT may be divided into sporadic and familial forms. Sporadic PAPT is charecterised by cerebellar dysfunction, visual disturbances and palatal tremors at 2Hz. Patients typically have olivary hypertrophy and increases signal intensity on T2-weighted MRI [7]. Hypertrophic olivary degeneration [HOD] is a transsynaptic degeneration of the inferior olivary nucleus of the medulla because of a lesion within the boundary of the GM triangle [8,9]. For many sporadic PAPT cases, a neurodegenerative aetiology is postulated however, some patients with structural brain lesions or autoimmune diseases were reported to develop PAPT as well [10]. In sporadic PAPT, the most prominent feature is progressive cerebellar degeneration, presenting as a poorly understood neurodegenerative disorder with onset in mid to late adulthood [11]. Familial PAPT is more complex than sporadic PAPT and may result from various etiologies including Alexander's disease, polymerase gamma mutations, spinocerebellar ataxia type 20, and GM2 gangliosidosis [11]. Familial PAPT differs from sporadic PAPT in having marked atrophy of cervical cord and brainstem with corticospinal signs, although the hypertrophic olivary appearance on MRI may be absent.

In our case patient presented with progressive ataxia, palatal tremors and other cerebellar features that aligns with syndrome of progressive ataxia with palatal tremors. With some significant family history. Patient has no confounding history of trauma, stroke, surgery. Radiological findings revealed degenerative changes in brainstem with corticospinal tract degeneration with features suggestive of familial PAPT and also had cerebellar degenerative changes.

However considering features like spasticity in all limbs with exaggerated deep tendon reflexes suggestive of corticospinal tract involvement, and with some significant positive family history and inconclusive genetic testing were suggestive with diagnosis of familial progressive ataxia with palatal tremor (PAPT).

In our patient, after a few days of treatment frequency of palatal tremors decreased from 140 - 160 range to 90 to 110 range. However there is no improvement in ataxia. Patient is on regular follow up. There is no known effective treatment for the progressive ataxia, which is the most disabling symptoms of PAPT [2]. Further studies and trails are required to potentially provide more targeted therapies.

This case adds to the limited literature on familial PAPT, particularly in the context of differentiating familial and sporadic forms with inconclusive genetic results.

Conclusion

This case report highlights the importance of clinico-radiological correlation while differentiating familial and sporadic forms of PAPT. Knowledge of PAPT is very important in clinical practice in further evaluation of patients presenting with palatal tremors. It is also important to know how to differentiate sporadic PAPT from familial with clinical feature and MRI features even when genetic test is inconclusive.

Conflict of Interest

None.

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