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Case Report

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Paroxysmal Upgaze in an Adult; Not always a Seizure, a Case Report

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ABSTRACT

Background: Paroxysmal Tonic Upgaze (PTU) is a rare neuro-ophthalmological syndrome typically observed in infancy or early childhood. Adult-onset and recurrent forms are exceedingly uncommon and may mimic other neurological or functional disorders, making diagnosis particularly challenging.

Case Presentation: We report a rare case of a 20-year-old male presenting with recurrent episodes of sustained upward eye deviation, preserved consciousness, and spontaneous resolution after sleep Seizures, fever, or neurological impairments were not linked to the events. The brain MRI, EEG, and neurological evaluation were all normal. Interestingly, the patient's sister's comparable experience raises the possibility of a family susceptibility. The clinical picture was in line with idiopathic, familial PTU, even though genetic testing was not widely available.

Discussion: This case illustrates the diagnostic difficulty of adult-onset PTU due to its rare occurrence and resemblance to conditions like seizures and oculogyric crisis, underscoring the need to recognize its benign course and characteristic features to avoid misdiagnosis and unnecessary treatment.

Conclusion: Adult-onset, familial PTU is an exceptionally rare entity. Increased awareness among clinicians is crucial for timely recognition and management. Further research is warranted to explore the genetic basis and spectrum of atypical PTU presentations.

Introduction

Paroxysmal tonic upgaze (PTU) is a rare, usually pediatric-onset, neuro-ophthalmologic condition characterized by episodic upward deviation of the eyes with preserved consciousness. Adult-onset is very rare.

The rare disorder known as Paroxysmal tonic upgaze (PTU) was initially identified by Ouvrier and Billson in 1988 as a separate clinical entity [1]. It is an age-related neuro-opthalmologic condition that mostly manifests in early childhood or infancy. Adult-onset is very rare.

It is defined by periods of prolonged tonic conjugate upward deviation of the eyes, frequently accompanied by horizontal eye movements that appear normal and down-beating saccades during attempted downgaze, often with compensatory chin-down head posture and preserved consciousness. Sleep usually relieves these symptoms where as these manifestations are frequently exaceberated by exhaustion, concurrent infections, or vaccinations. Other neurological symptoms like ataxia, psychomotor retardation, intellectual disability, nystagmus, amblyopia, or strabismus have been linked to this illness in certain studies [2].

Symptoms typically begin in early infancy, improve during sleep, and often resolve spontaneously. However, secondary causes must be ruled out. Diagnosis is challenging for clinicians due to overlap with conditions like dystonia, seizures, and non-epileptic events. Video EEG can aid diagnosis, though it's usually normal, with occasional occipital discharges. Brain imaging is generally unremarkable, although occasional abnormalities in the visual pathway have been reported [3].

The pathophysiology of PTU is not clearly understood: genetic susceptibility, immunological factors, dorsal brain stem immaturity, and neurotransmitter depletion are thought to be the

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causes. Until now, mutations in three genes, CACNA1A, GRID2 and SEPSECS have been associated with PTU [4].

The patients' laboratory results, cerebral imaging, and electroencephalogram (EEG) are all normal. Nonetheless, some research recommend additional testing for individuals exhibiting neurological symptoms such nystagmus, developmental delay, and abnormal magnetic resonance imaging [5]. Most cases resolve by early childhood. Adult-onset and recurrent forms are very rare and may mimic seizure activity, oculogyric crises, or functional disorders.

We present a rare case of adult-onset, recurrent PTU with 3 documented episodes over four years. Voluntary downward movement of eye during episodes and complete recovery after sleep were is a distinguishing clinical feature.

This case expands the understanding of recurrent, adult-onset PTU and reinforces its largely benign nature.

Case Presentation

A 20 years old male with NKCM, presented to neurology OPD with complaint of a prolonged 12-hours episode of sustained involuntary tonic upgaze, interrupted by multiple voluntary incomplete downward saccades during the episode. During this episode, he remained fully conscious and oriented with no associated fever, hallucinations, visual field loss, seizures, or limb involvement.

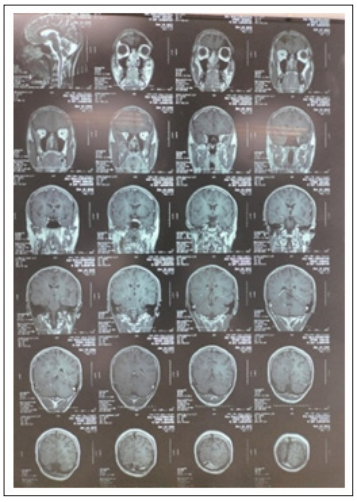
Patient went to the emergency department of a tertiary care hospital and was admitted. Neurological examination was normal apart from sustained upward gaze. IV paracetamol was administered. The episode resolved spontaneously after he fell asleep, and he woke up with normal eye position.

He reported two similar episodes of almost same duration, one occurred 2 years ago and the other one 4 years ago, which also resolved spontaneously after waking up from sleep.

Patient also reported similar episodes in his biological sister which suggests a familial or genetic predisposition.

Neurological examination was unremarkable. Ophthalmology assessment showed visual acuity of 6/6 with normal pupillary reflexes and fundus. There was no diplopia, nystagmus or ophthalmoplegia.

MRI and EEG both were also unremarkable.



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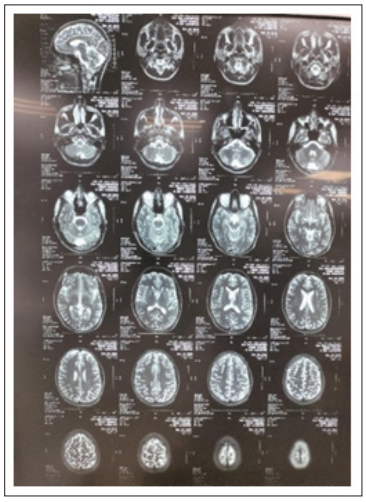

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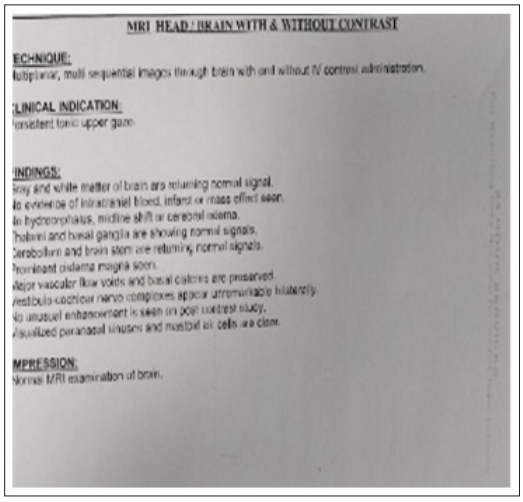
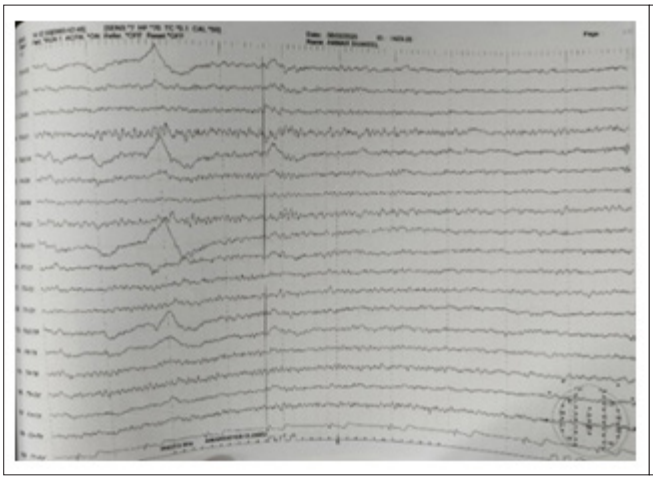


Figure: 3

Figure 1, 2 and 3: Normal Brain Mri Findings



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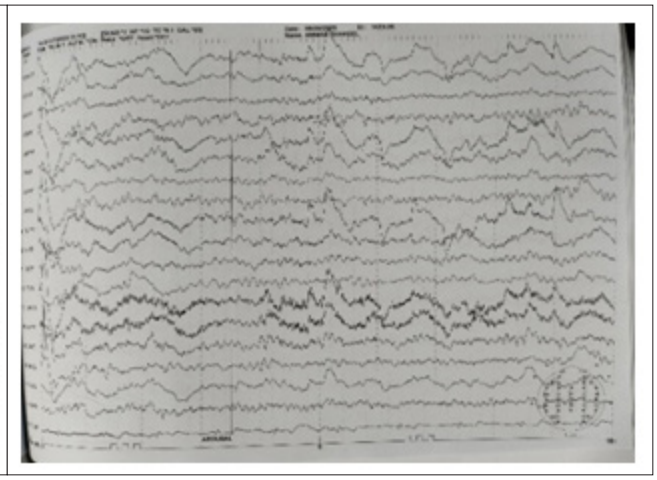

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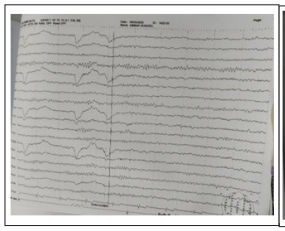

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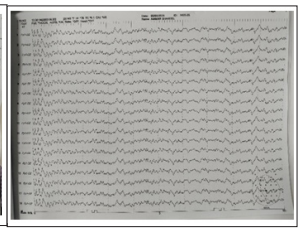

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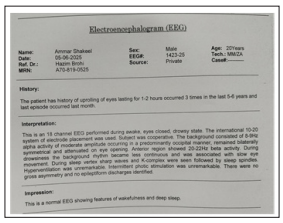

Figure: 8

Figure 4,5,6,7 and 8: EEG Findings Showing No Abnormal Activity

Discussion

Paroxysmal Tonic Upgaze (PTU) is a rare neuro-ophthalmological disorder that primarily affects infants and young children, with adult-onset cases being exceptionally uncommon and rarely documented. It presents as recurrent episodes of sustained, conjugate upward eye deviation, while horizontal eye movements and consciousness remain intact. First described by Ouvrier and Billson in 1988, most cases demonstrate early childhood onset, spontaneous remission, and a typically benign course [1].

The 20-year-old male patient we treated had the usual symptoms of PTU: intact awareness, spontaneous resolution during sleep, episodic tonic upward gaze, and no neurological or systemic abnormalities. However, this case is clinically significant since adult-onset PTU is extremely uncommon, especially when it recurs over years. A case report by Lalani et al. (2021) reported PTU as one of the paroxysmal movement symptoms in a pediatric patient with SCN8A-related developmental and epileptic encephalopathy, showing that PTU can arise after infancy and in complex genetic disorders [6]. Although not an adult-onset example, it broadens the clinical spectrum of PTU presentations and emphasizes the importance of considering genetic and age-atypical signs in diagnosis work up.

Our patient's voluntary partial downward saccades during episodes are a distinctive and unusual characteristic that is rarely reported in pediatric cases. This implies that partial oculomotor control is maintained, which can assist in distinguishing PTU from seizure activity or oculogyric crises, in which volitional movement is typically compromised.

Another noteworthy feature of this case is the positive family history, as the patient's sister reportedly experienced similar episodes. This raises the possibility of a familial or genetic form of PTU. While most cases are sporadic, mutations in CACNA1A, GRID2, and SEPSECS genes have been associated with hereditary PTU, indicating a genetic predisposition [4]. Although genetic testing was not feasible due to resource constraints, the recurrent yet benign course and familial clustering are consistent with patterns described in hereditary PTU cases.

Neuroimaging and EEG findings in PTU are generally normal, as was the case here. However, some studies have described transient occipital discharges on EEG or brainstem lesions on MRI [7]. mentions a case of 6 month old child with occipital discharge on electroencephalography (EEG) making the diagnosis more complicated [7].

PTU can resemble a number of other illnesses, such as brainstem pathology, oculogyric crisis, focal seizures (e.g., frontal eye field origin), and functional movement disorders. Careful clinical correlation,a standard investigative work up, and the identification of distinguishing characteristics (such as retained consciousness, sleep-induced resolution, and the lack of systemic signs) are necessary to distinguish between these.

This case highlights an adult-onset, recurrent form of PTU, with three documented episodes over a span of four years. The patient's ability to voluntarily bring his eyes downward during the episodes and complete resolution with sleep argue against seizure or dystonic crisis. The familial occurrence raises the possibility of a genetic or inherited ion channel dysfunction.

Although childhood PTU is often linked to channelopathies or cerebellar dysfunction, the etiology of adult-onset cases remains unclear.

While most pediatric cases resolve within a few years, the recurrent nature in adulthood, as seen in our patient, challenges the notion of PTU as an exclusively pediatric and self-limited condition. This instance emphasizes the necessity of raising knowledge among adult neurologists and ophthalmologists in order to prevent incorrect diagnoses and unnecessary interventions.

Conclusion

This case illustrates a rare occurrence of recurring, adult-onset paroxysmal tonic upgaze (PTU) in a healthy individual with a possible familial predisposition. The diagnosis of idiopathic PTU is supported by the patient's characteristic clinical symptoms, which include episodic tonic upgaze, retained awareness, spontaneous resolution with sleep, and a normal neurological workup. The absence of identifiable structural or epileptic pathology reinforces the benign nature of this condition. Given the rarity of adult-onset PTU and its overlap with seizure and other movement disorders, PTU poses a diagnostic challenge for neurologist, often leading to misdiagnosis. Therefore, increased clinical awareness is essential to avoid unnecessary investigations.

Understanding the pathophysiology and inherited patterns of PTU may be improved with the use of genetic analysis and additional documentation of similar instances that are comparable. Hence, further research is needed to better understand the pathophysiology, genetic basis, and spectrum of atypical presentations in PTU, particularly in adult-onset and familial cases.

Consent for publication

Informed consent was taken from the patient for publication.

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