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Case Report

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PAROXYSMAL TONIC UPGAZE MIMICKING INFANTILE SEIZURE

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Abstract

Non-epileptic paroxysmal events are very important in daily clinical practice. Paroxysmal tonic upgaze is a rare neuro-ophthalmologic non-epileptic paroxysmal condition characterized by episodes of continuous conjugated upward deviation of the eyes and normal horizontal gaze. Its pathogenesis is unknown, and its etiology is heterogeneous. The prognosis is often benign, but it's often confused with epilepsy. We present a case of "paroxysmal tonic upgaze" with a benign clinical course and spontaneous remission.

Keywords: Pediatric, non-epileptic, paroxysmal tonic upgaze.

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Introduction

Paroxysmal tonic upgaze (PTU) of childhood is a very rare neuro-ophthalmologic syndrome characterized by short-term episodes of upward deviation of the eyes. The age of onset varies between 1 month and two years. PTU is characterized by conjugated upward gaze attacks and accompanying neck flexion (chin down) to compensate for upward gaze. [1,2]

The diurnal course of the attacks and their disappearance during sleep, and the preservation of consciousness are very important for the differential diagnosis of epilepsy. In some cases, ataxia and developmental delay may accompany them. The pathophysiology of PTU is not clearly understood: genetic predisposition, immunological causes, dorsal brain stem immaturity, and depletion of neurotransmitters are considered as reasons.[1,3] Electroencephalography (EEG), cranial imaging and laboratory tests of the patients are normal. However, some studies suggest further examinations for patients with neurological symptoms such as developmental delay, nystagmus, and abnormal magnetic resonance imaging.[2,4,5]

In this article, we aimed to present a case of PTU, which is a rare, benign condition with a post-infectious cause.

Case Report

A 6-month-old female patient applied to the health institution with recurrent attacks accompanied by a sudden upward gaze in the eyes and neck flexion, which started two weeks after treatment for the diagnosis of bronchiolitis due to cough and fever complaints that started 20 days ago (Figure 1). She was referred to our clinic with the suspicion of epilepsy, as the attacks continued increasingly during the day. It was stated that the patient was full-term born with a normal delivery as the first child of the family. A history of phototherapy due to jaundice in the neonatal period was stated in the history. The patient's mental motor development was normal. Her parents were not relatives, and her cousin had a diagnosis of epilepsy.

The episodes, which did not exceed 20 seconds, started two days before her application, and their frequency has gradually increased. There was no change in consciousness during the attacks, and it was described that these attacks disappeared during sleep. On physical examination of the patient, growth parameters were in the normal percentile range. Neurological examination, including cranial nerves, ophthalmological examination and other system examinations, were normal. She had no nystagmus or ataxia during the attacks. Complete blood count, thyroid function tests, vitamin B12, folate, iron, iron-binding capacity and biochemistry parameters were normal. No seropositivity was detected in the viral panel. EEG and brain magnetic resonance studies were normal. During the follow-up, the number and duration of the attacks decreased without

treatment and decreased significantly after the seventh day. Attacks were rarely described in the first month of follow-up.

Figure 1. Upward gaze in the eyes of the patient at the time of admission.

Discussion

Paroxysmal tonic upgaze of childhood was first described as a benign disease by Ouvrier and Billson in 1988. The age of onset of PTU ranges from 1 week to 90 months. [1,3,4] Our patient was six month-old and had normal neuromotor development.

Although the pathophysiology is not clearly known, the condition is often not observed by means of tests such as physical examination, neuroimaging, and EEG. It is thought that conditions such as mutations like CACNA1A, immunological events such as vaccination and inflammatory diseases, vitamin B12 deficiency and immaturity of the central nervous system alone or together cause PTU. It has been reported that attacks increase in stressful conditions such as inflammatory disease, fatigue, and constipation. [2,5,6] In addition, the duration and frequency of attacks also vary. The attacks gradually decrease and disappear, and Verrotti et al. reported in their study that tonic upgaze attacks disappeared between 1 and 4 years without any treatment and without any change in psychomotor development. Attacks may increase in cases of stress, such as inflammatory diseases, insomnia, and fatigue. It is seen that many patients recover spontaneously at preschool age without the need for treatment and without causing any neuromotor developmental delay. However, mild learning difficulties were reported in some cases, and severe learning difficulties were reported in up to 40%. [4,7] In our patient, the attacks started after bronchiolitis and varied during the day. Her neuromotor development was

normal, and EEG and brain MRI showed no abnormalities. The complaints disappeared with sleep and regressed over time without treatment. Neuromotor development was normal.

Some treatment regimens, such as antiepileptic agents and levodopa, have been suggested. A noticeable recovery has been noted in some patients using levodopa, but the benefit of antiepileptic drugs has not been demonstrated. However, levodopa therapy is not routinely recommended. In patients with low vitamin B12 levels, regression has been reported in complaints with vitamin B12 supplementation. It has been demonstrated that carbonic anhydrase inhibitors are effective in patients with CACNA1A mutations.[4,6-8] Recovery was observed in our patient without the need for any treatment, and it was observed that the complaints disappeared with the regression of the immunological status.

Since there was no structural lesion or any cause, such as vaccination, the patient we presented was considered a post-infectious paroxysmal tonic upgaze. In conclusion, PTU is a transient, benign condition that is extremely rare in childhood. It is very important since epilepsy is included in the differential diagnosis and is rarely seen.

Ethical Considerations: Informed consent has been obtained from the parents of the patient.

Conflict of Interest: The authors declare no conflict of interest.

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