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Benign paroxysmal torticollis of infancy

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Abstract

Benign paroxysmal torticollis of infancy (BPTI) is a rare self-limiting disorder characterized by recurrent episodes of head tilting accompanied by periods of irritability, ataxia, emesis and weakness. It occurs within the first few months of life, it usually improves by 2 years and typically resolves by 3 years. It is often underdiagnosed and leads to unnecessary extensive workup. We describe a case of an 11- month-old infant with BPTI.

Keywords

BPTI; emesis; weakness; infants

Introduction

Benign paroxysmal torticollis of infancy (BPTI) is an underdiagnosed condition, and only a few cases have been reported in the literature [1,2,3,4]. If not considered in the differential diagnosis, these infants may undergo extensive neurological work up. Therefore, it is important to recognize this condition early to avoid unnecessary medical work up and alleviate parental anxiety. Most cases present in early infancy and rarely during late infancy. We present an 11-month old infant with BPTI.

Case Report

An 11-month-old Caucasian female presented to emergency department with emesis, weakness in upper extremities and torso, accompanied by right sided torticollis. Her symptoms started 1 day prior to admission, with sudden onset of weakness in extremities and torso causing her difficulty in crawling and imbalance while sitting. She had 6-7 episodes of emesis. Mother reported she was irritable and crying.

Past medical history is significant for intermittent torticollis that started at 4-5 months of age. Parents reported she would have right sided torticollis for few weeks, left side for few weeks, and sometimes she can be normal without torticollis. Three months prior to current admission, she had a similar episode of the weakness of torso and emesis. She was seen in the emergency department at that time, had lab work done, that was normal. She also had a computed tomography scan of the head done which was normal. Her symptoms improved in 6-8 hours and the patient was sent home. Family history is significant for severe migraines in father.

On physical exam, she had right-sided torticollis and difficulty supporting her head and torso

while sitting. A complete blood count, serum electrolytes, C-reactive protein, creatine kinase, and serum cortisol were normal. The patient was given intravenous fluid and within 4-5 hours her weakness improved and her emesis was controlled. She was discharged home after a brief period of observation.

Discussion

BPTI is a self-limiting paroxysmal condition, often under diagnosed. It typically occurs in the first few months of life, improves by 2 years of age and generally resolved by 3 years of age [1-4]. BPTI is currently classified as one of the childhood episodic syndromes. Other conditions classified as childhood episodic syndromes include, abdominal migraine and cyclic vomiting syndrome, and considered as common precursors of migraine [5]. Based on the duration of symptoms BPTI can be classified into two types, 1) periodic torticollis (episodes last for several hours or days), and 2) paroxysmal torticollis (episodes last for a few minutes and are accompanied by ocular signs) [1-4].It is most commonly reported in females, with a male to female ratio of 3:1 [6]. It often presents as early morning retrocollis, later torticollis, often triggered by postural changes. The etiology of BPTI is unknown, but it is postulated to be due to disorders in the central vestibular region or vestibulocerebellar connections, especially when ataxia is associated. It is known to be associated with CACNA1A mutation if there is a strong family history of familial hemiplegia, episodic ataxia, paroxysmal tonic upgaze [7,8,9,10]. One study using positron emission tomography showed a decrease in glucose metabolism in the basal ganglia and the cerebellum [4].

Neurologic exam between attacks, electroencephalogram and magnetic resonance imaging scan done are normal. No work up in indicated if a good history and examination is done which gives away the diagnosis. BPTI has a strong correlation with migraines and can be associated with neurodevelopmental delay [5]. A national, telephone survey of pediatricians, showed only two out of 82 (2.4%) pediatricians stated that they were aware of the condition telling us that BPTI is underdiagnosed [1].

No medication has shown proven benefit so far. Supportive care with fluids and anti-emetics is sufficient. Some studies trialed cyproheptadine with some benefit, however safe dose for children younger than 2 years is not studied. A trial of topiramate at a dose of 2 mg/kg was tried in four patients, given correlation of BPTI with migraines, which has shown improvement without side effects [6].

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