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Case Report

Benign Paroxysmal Torticollis of Infancy: Has it Outgrown its Definition?

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Abstract

Benign Paroxysmal Torticollis of Infancy (BPTI) is considered a migraine variant and a precursor of other migraine symptoms. It is classified as a condition that starts before one year of age with symptoms resolving by the age of five. We describe two unrelated children over the age of five who presented with features of BPTI. We would propose that the definition of the condition is therefore extended beyond infancy, and it is possible, if not likely, given the rarity and obscurity of the condition, that it may be overlooked in children of all ages.

Keywords: Paroxysmal; Torticollis; Channelopathies; Migraine

Abbreviations

BPTI: Benign Paroxysmal Torticollis of Infancy; CD: Cervical Dystonia.

Introduction

Benign Paroxysmal Torticollis of Infancy (BPTI) is a well described if not well recognized [1] condition characterised by episodes of torticollis which occur spontaneously and last between half an hour and a few days, before complete spontaneous resolution. The torticollis can be painful and there may be accompanying symptoms such as vomiting, drowsiness, irritability and ataxia. Neurological examination between episodes is normal and although there may be an association with slow development, BPTI does not appear to have long term sequelae [2].

BPTI is of unknown aetiology but has many features suggestive of a channelopathy. It is usually described as a migraine precursor with a possible genetic link to hemiplegic migraine [3-5], however many patients do not proceed to migraine in later life [6]. Of note, as its name implies, it has only been described in children under the age of five.

We describe two children over the age of five, who presented with features of BPTI within a few months of each other to a District General Hospital in Hertfordshire UK. We would propose that the definition of the condition is therefore extended beyond infancy, and it is possible, if not likely, given the rarity and obscurity of the condition, that it may be overlooked in children of all ages.

Case 1

An 11-year-old Caucasian girl presented with a 2-month history of constipation and was admitted under the paediatric team for a disempaction regime. A full past medical history was taken which revealed an unrelated history of episodes of severe "neck stiffness". Initially these episodes had been fleeting and had been ascribed to poor posture but with time they became more intense and more frequent. They were now occurring every few months and lasting days to weeks with severe episodes lasting 6-8 weeks. Initially the parents thought that these episodes had started a few years ago, however in hindsight they realized that she may have been having milder attacks since young infancy.

At the time of presentation each episode was immensely painful and associated with spasms affecting various areas of the neck such that there could be associated tilting to one side or the other, or forward flexion. There were no obvious precipitants and no history of trauma. At these times her parents described the neck muscles as 'wood hard'. Onset would be sudden, but her eyes appeared 'glazed' prior to an attack.

She had attended the Emergency Department twice previously during serious episodes and had been prescribed simple analgesics, diazepam and hot or cold compresses to little effect. Recovery would begin with the muscles relaxing and the symptoms would then subside over 2-3 days. In between episodes she was entirely well and able to participate in all activities.

There was no personal history of classical migraine although she sometimes suffered with a headache secondary to the neck

pain. In addition to this she had longstanding constipation, mild eczema and had an adenotonsillectomy aged 4 years. Two older siblings aged 18 and 22 both suffered with migraine as did both Paternal and Maternal grandmothers.

During her admission for disimpaction – despite her age – a presumptive diagnosis of BPTI was made and she was started on Pizotifen 0.5mg daily leading to an improvement in the severity of symptoms and longer symptom free periods. She did nonetheless have one further major attack and the dose was increased to 1mg daily. An MRI of her brain was completely normal.

She was reviewed by a specialist paediatric neurology team once she was established on Pizotifen 1mg at night. They concurred with the diagnoses of BPTI and recommended continuing medication at the current dose. A plan for further investigations was made including DYT-1 analysis and referral to a movement disorder team if further relapses were to occur. However, she had had no significant episodes in the 3 years since starting the higher dose of pizotifen. She felt reassured by the pizotifen and was reluctant to stop it. She was eventually convinced to stop taking pizotifen and has been free of episodes for 3 months at the time of writing.

Case 2

A nine-year-old girl Caucasian girl was seen referred acutely to the Paediatric Team with severe right sided torticollis. This was her third episode in two years. Each had been very painful and had resulted in an emergency department presentation. Each episode had been treated with simple analgesics and 'responded' completely.

This episode, just like the previous two, had come on suddenly with no warning and no obvious trigger. There had been no history of trauma and she was entirely well between episodes. She had tried simple analgesics and diazepam to little effect. Episodes lasted between 2 and 7 days and left her incapacitated due to the pain. During this episode her head was fixed in the right lateral position and even the slightest movement was excruciatingly painful.

There was no personal history of classical migraine but a strong family history with her mother, maternal aunt and maternal grandmother all suffering with migraine.

She was discharged home from the emergency department and followed up 2 days later on the Day Admission Unit. An MRI brain and cervical spine showed no abnormalities. Routine bloods were normal. There was a strong suspicion of this being a variant of BPTI and hence she was given a trial of pizotifen and referred to physiotherapy.

She was followed up in clinic where she reported no relief from the exercises given by physiotherapist, but she had also seen an osteopath privately and felt there was a good response to this. She stopped taking Pizotifen as this made her too sleepy. She had no further episodes for 18 months however she had gone that long between episodes previously.

Discussion

BPTI is a rare condition and this is likely to contribute to its presumed underdiagnosis. It is recognized by the International Headache Classification of Headache Disorders (ICHD)-III [7] as one of the "episodic syndromes that may be associated with migraine". By definition it starts within the first year of life with paroxysmal episodes of torticollis which last from a few minutes to a week. There can be associated symptoms such as vomiting, pallor, malaise, irritability and in older children ataxia. The episodes recur on a regular, often monthly basis. They tend to improve such that they have been described as terminating by the age of five [8]. The current belief is that BPTI is a likely migraine precursor with many patients progressing to display typical migraines in later life and often presenting with a strong family history of migraine.

In adults similar symptoms are described in cervical dystonia (CD). Onset is usually over 40 years of age and although episodes of dystonia may cause headache there is no clear link between CD and migraine. Given the personal and family histories of the children described in this report they fit much more comfortably with a diagnosis of BPTI than they would with one of CD.

With increasing knowledge there is a recognition that migraine and its equivalents may have multiple causes beyond a single vascular aetiology. It is thought that BPTI may be included in channelopathy–related genetic syndromes which include familial hemiplegic migraine, paroxysmal upgaze and episodic ataxia [9]. These seem to have a common link with CACNA1A mutations which may influence calcium channels [2-4].

Conclusion

We present two cases which fit the clinical criteria of BPTI. Both have occurred considerably beyond the age at which these attacks are said to subside, and at least one commenced when the child was out of infancy. Given that there are less than a few hundred cases described in the world literature, it is a coincidence that two presented within a short period time to a general paediatric service. This makes it likely that many cases present elsewhere but may be unrecognized. We hope that this report serves to raise the awareness of this condition in doctors who treat children. Further we would propose expanding the diagnosis to extend beyond infancy as this would allow the thinking clinician to identify the problem when it does arise in the older patient. We should let BPTI grow out of its infancy and at least enter later childhood.

Conflict of Interest

The authors have no conflicts of interest relevant to this article to disclose.

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