CASE REPORT

Benign paroxysmal torticollis in infancy: Report of two cases and aspects on pathogenesis

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ABSTRACT

Benign paroxysmal torticollis in infancy is characterized by periods of torticollis, usually occurring at the first year of life, at varying time intervals and typically resolves without complications within 1-5 years. It is thought to be a benign, self-limited paroxysmal disorder and has been regarded as a migraine equivalent, but the precise etiology remains unknown. We report two cases of recurrent episodes of torticollis and theories of etiology and pathophysiology are discussed including the potential role of mutations in the CACNA1A calcium channel.

Key words: *benign paroxysmal torticollis, migraine equivalents, movement disorders*

BACKGROUND

Benign paroxysmal torticollis in infancy (BPTI) was first described by Snyder in 1969.[1] This condition is characterized by recurrent episodes of torticollis, sometimes followed by vomiting, pallor, ataxia, irritability or drowsiness, which resolve spontaneously within a few hours or days.

The pathogenesis of this disorder remains unknown. Some researchers regard BPTI as a migraine equivalent of infancy.[2,3] It has also been reported that some patients with BPTI develop paroxysmal vertigo or migraine at an older age. Recent studies suggest that BPTI, benign paroxysmal vertigo and migraine are different phenotypic expressions of the same calcium channelopathy.[2]

We present two infants with recurrent episodes of BPTI and report the follow-up data to the outcome of the condition. We have also conducted a review of the literature on the pathogenesis of the disorder.

PRESENTATION OF TWO CASES

The first patient was an 8-month-old girl) who was brought to the pediatric neurologic outpatient clinic due to recurrent episodes of torticollis from the age of 5 months. The episodes were usually noted upon awakening in the morning, alternated from side to side, lasted approximately 7 days and recurred after 7 days. The episodes resolved spontaneously with no residual deficit in the intervals. No other acute symptoms were associated with the paroxysmal torticollis in this case.

The infant was the second child of apparently healthy parents. The father of the infant suffered from recurrent episodes of headaches, not further investigated. The patient had a healthy 2-year-old sister. She was born after an uncomplicated pregnancy (planned cesarean section), with a birth weight of 2850 gr. The patient had a normal physical and psychomotor development. She had received all the doses of the routine childhood immunization schedule for her age. Serious medical problems or use of medications were not reported in the infant's history. Symptoms of gastroesophageal reflux were not noted.

Painless torticollis on the right side was diagnosed on physical examination. The standardized neurologic evaluation and the rest unremarkable physical findings did not support the diagnosis of a neurologic deficit.

The precise choice of laboratory tests required for an evaluation of paroxysmal torticollis has not been standardized but can be seen as two sets. In the first set are those studies needed for evaluation of secondary diseases that could mimic a BPTI. These studies include a complete blood count, erythrocyte sedimentation rate(ESR), screening chemistries of liver and kidney function, and blood glucose levels, as well as C-reactive protein (CRP).

Laboratory investigation revealed the following results: WBC count 17000 /mm^3 with a predominance of lymphocytes, Hb(Hemoglobin) 11,7 g/dl, Ht 35%, ESR 11 mm/ 1h, CRP 3,5 mg/L and normal biochemical profile. The second area of laboratory testing relates to the detection of associated disorders.Orthopedic abnormalities were excluded with conventional radiography of the cervical spine. Ultrasonography of the brain and neck were also performed, with normal findings. The sonographic appearance of the gastroeophageal junction and the 24- hours pH monitoring of the oesophagus did not indicate the presence of gastroesophageal reflux. MRI of the brain and EEG(electroencephalogram) as well as the detailed ophthalmologic and ENT (Otolaryngologic) tests within physiologic patterns.

The infant was followed-up up until 13 months of age and continued to have recurrent episodes of torticollis with the same clinical characteristics and without the development of secondary symptoms or/and physical findings.

The second case involves an 8-month-old girl who presenting with torticollis, pallor, localized cyanosis around the mouth- lasting for a few seconds only- vomiting and drowsiness. Three previous episodes of paroxysmal torticollis were noted in her history, which had resolved after paracetamol administration, according to the parents. No residual abnormality was observed during the intervals.

The infant was the first child of apparently healthy parents. She was born by cesarean section at the 38th week of

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