SHORT COMMUNICATION

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Benign paroxysmal torticollis in infancy – diagnostic error possibility

Benigni paroksizmalni tortikolis u detinjstvu – mogućnost dijagnostičke greške

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Abstract

Background/Aim. Benign paroxysmal torticollis (BPT) is a rare paroxysmal dyskinesia characterized by attacks of head tilt alone or tilt accompanied by vomiting and ataxia, which may last hours to days. It is claimed that BPT disappears completely in childhood, but that it can evolve into other conditions, such as benign paroxysmal vertigo, cyclical vomiting syndrome, abdominal migraine, hemiplegic migraine, motion sickness and/or migraine with aura. The aim of this manuscript was to renew focus on benign paroxysmal torticollis because the disorder is almost always underrecognized by pediatric practitioners, who often order extensive and unrewarding testing and physiotherapy treatment. **Methods.** Twelve BPT cases observed during a 5-year period (2009–2014) at the Clinical Centre Niš, Niš, Serbia were reviewed. Data were collected on the features of torticollis, the age of onset, the duration of episodes, associated symptoms, the frequency of episodes, the persistence of symptoms over time, the age when the disorder finally disappeared, sequelae appearing after the 5th birthday, and family history of BPT, migraine or kinetosis. All the children were followed for periods ranging from 48 to 72 months. **Results.** The series included 6 females and 6 males. The age at onset of BPT was less than 8 months in 84% of the cases. Episodes of torticollis occurred suddenly on waking in all the cases without any trigger factors. The duration of torticollis ranged from a few hours to a few weeks. In 58% of cases, the condition persisted for more than one week. The frequency of the episodes ranged from once every 3 days to once every 25 days. The episodes were more frequent and lasted longer in the early months and tended to cease as the child became older. The age when episodes ended ranged from 11 months to 62 months. In 11 (91.66%) cases, the disorder disappeared before the patient's 5th birthday. No patient had a family history of BPT. In 6 cases, family members had kinetosis. In 5 cases, family members were positive for both migraine and kinetosis. All the children had normal motor development and normal speech and language development. After the disappearance of BPT, two children developed other forms of periodic syndromes: one boy had migraine with aura, and one girl experienced cyclic vomiting. **Conclusion.** BPT is probably an age-sensitive and migraine-related disorder that is benign in nature. The disorder is often misinterpreted, and children may pointlessly undergo numerous tests. Therefore, it is very important to recognize and observe this condition in order to avoid extensive, unnecessary and unpleasant procedures on the child.

Key words:
torticollis; diagnosis; diagnostic errors; child.

Apstrakt

Uvod/Cilj. Benigni parosizmalni tortikolis (BPT) je retka paroksizmalna diskinezija koja se manifestuje iznenadnom pojavom prinudnog položaja glave, izolovano ili udruženo sa povraćanjem i ataksijom. Smatra se da BPT kompletno iščezava u detinjstvu ili evoluira u druga stanja kao što su: benigni paroksizmalni vertigo, sindrom cikličnog povraćanja, abdominalna migrena, hemiplegična migrena, poremećaj pokretljivosti i/ili migrena sa aurom. Cilj ovog rada bio je da se fokus usmeri na BPT, jer ovaj entitet često ostaje neprepoznat u pedijatrijskoj praksi, pa se preduzimaju opsežne i nepotrebne dijagnostičke i terapijske procedure. **Metode.** Tokom petogodišnjeg perioda (2009–2014) u Kliničkom centru Niš, (Niš, Srbija), praćeno je dvanaestoro dece sa BPT. Prikupljani su podaci o obeležjima tortikolisa, starosti dece prilikom inicijalne pojave poremećaja, dužini epizoda i pridruženim simptomima, o učestalosti epizoda, starosti u kojoj je poremećaj iščezao, posledicama prisutnim nakon petog rođendana i podacima u vezi sa porodičnom anamnezom BPT, migrene i kinetoze. Sva deca su praćena tokom perioda od 48–72 meseca**. Rezultati.** Praćenjem je obuhvaćeno 6 dečaka i 6 devojčica. Prva pojava simptoma je kod 84% ispitanika bila pre 8. meseca života. Kod svih ispitanika epizode tortikolisa javljale su se iznenada i spontano prilikom buđenja. Tortikolis se ispoljavao u trajanju od

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nekoliko časova do nekoliko nedelja. Kod 58% ispitanika tortikolis se održavao više od jedne nedelje. Frekventnost epizoda kretala se od jednog pojavljivanja u tri dana do jednog pojavljivanja u 25 dana. Epizode su bile češće i duže su trajale u prvim mesecima života i imale su tendenciju prestanka sa odrastanjem deteta. Uzrast kada su se epizode sa tortikolisom završavale kretao se u rasponu od 11 do 62 meseca. Kod 11 (96%) ispitanika poremećaj je nestajao pre petog rođendana. Kod svih ispitanika nije postojala pozitivna porodična anamneza za BPT. Kod 6 ispitanika članovi porodice su imali kinetozu, dok je kod 6 ispitanika porodična anamneza bila pozitivna i za migrenu i kinetozu. Svi ispitanici su imali normalan razvoj motoričkih funkcija i sposobnosti govora. Nakon nestanka BPT, kod dvoje dece razvili su se drugi oblici periodičnih sindroma. Kod jednog dečaka ustanovljena je migrena bez aure, dok su se kod jedne devojčice ispoljila ciklična povraćanja**. Zaključak**. BPT je benigni poremećaj vezan za rano detinjstvo i često udružen sa migrenom. Obično se ne dijagnostikuje na vreme, pa se deca bespotrebno podvrgavaju brojnim ispitivanjima. Zbog toga je veoma važno blagovremeno prepoznati i pratiti ovo stanje čime bi se izbegle obimne, nepotrebne i neprijatne dijagnostičke i terapijske procedure.

Ključne reči:
tortikolis; dijagnoza; dijagnostičke greške; deca.

Introduction

Benign paroxysmal torticollis (BPT) is a rare paroxysmal dyskinesia characterized by attacks of head tilt alone or tilt accompanied by vomiting and ataxia, which may last hours to days [1]. It is an episodic functional disorder of unknown etiology that occurs in early infancy. It is characterized by recurrent episodes of abnormal rotation and inclination of the head to one side, which are sometimes accompanied by an asymmetric posture of the trunk with bending toward the same side (tortipelvis) [2]. Attacks can cause distress to the child and parents, but unfortunately, symptomatic treatment has not been helpful. The differential diagnosis may be difficult in case when the first attack occurs in a previously healthy infant. It includes seizures, vertigo, gastroesophageal reflux, idiopathic torsion dystonia, dystonic reactions to drugs, stroke, familiar hemiplegic migraine, alternating hemiplegia of childhood, ocular palsy, posterior fossa tumors, cervical spine abnormalities and Sandifer syndrome [3–5].

A study has been recently conducted with the aim of determining the general awereness of pediatricians of BPT. The study has shown that only 2 out of 82 pediatricians (2.4%) are aware of this condition [6].

The diagnosis of BPT in infancy is based on clinical grounds: a proper history and physical examination in paticular. Other examinations, such as neuroimaging, long-term video EEG recording or genetic studies, are necessary only if other diagnoses are suspected and other more serious conditions should be excluded.When the tests prompted by the first episode reveal negative findings, the parents relax and do not return to the pediatrician. The recurrent nature of the episodes is what leads to the diagnosis of BPT [2, 7].

Currently known facts about benign paroxysmal torticollis in infancy

There are relatively few reports on this disorder in the literature, with approximately 100 cases being reported since 1969. BPT was first described in 1969 by Snyder [8], who reported 12 cases with onset in infancy.

BPT is a self-limited condition that occurs in the first few months of life, usually between the age of 2 weeks and 5 months. The disorder is characterized by recurrent episodes of torticollis, which is often accompanied by vomiting, pallor, irritability, ataxia and drowsiness. Attacks can occur on either side and can last for hours or days, but most attacks last for less than a week. The episodes recur every few days to every few months. Improvement is seen by the age of 2 years and the episodes end by the age of 5. There is often a family history of migraine and motion sickness. BPT is a self-limited condition that usually does not respond to any kind of treatment. Sometimes, coordination problems, particularly gross motor difficulties, may accompany BPT, suggesting that the disorder is probably brain-based and maturational in nature [9].

It is crucial to recognise the condition and to reassure parents of its benign course and not to be misdiagnosed for other disorders.

BPT is not considered to be a frequent cause of torticollis. In 700 cases of primary torticollis (followed from 1970 to 1988), only 7 cases (1%) were due to BPT [10]. The etiopathogenesis of BPT is unknown. Some authors suggest an underlying vestibular disorder such as labyrinthitis [8]. Others claim the involvement of the central vestibular region or vestibulocerebellar connections [11]. It is also believed that the immaturity of the brain or a deficiency of certain neurotransmitters during a limited period of life is also involved.

The hypothesis of channelopathy has been raised. This entity has been recently linked to a mutation in the CACNA1A gene [12, 13]. Giffin et al. [12] suggested in 2002 that BPT was a migraine equivalent, related to familial hemiplegic migraine and connected to a CACNA1A mutation on chromosome 19. Recent studies have shown that heterogeneous paroxysmal disorders which include BPT are associated with PRRT2 gene mutation [14, 15].

Recognising the possible presence of a genetic defect in at least some cases of BPT, certain authors have suggested that BPT is a developmentally sensitive disorder associated with immaturity of the central nervous system.

Kimura and Nezu [16] reported on findings related to surface electromyography (s-EMG) in one case with BPT and suggested that BPT should be categorized as an idiopathic paroxysmal dystonia in infancy. The common finding of the family history of migraine and/or kinetosis supports the hypothesis that BPT is caused in such patients not only by an immaturity of the neuronal system, but also by a hereditary

predisposition for persistent functional vestibular dysfunction [17, 18]. Other authors reported a positive family history of migraine and kinetosis in 25–55% of cases [2, 18, 19].

It is claimed that BPT disappears completely in childhood, but that it can evolve into other conditions, such as benign paroxysmal vertigo, cyclical vomiting syndrome, abdominal migraine, hemiplegic migraine, motion sickness and/or migraine with aura [20, 21].

The aim of this manuscript was to renew focus on benign paroxysmal torticollis because the disorder is almost always under-recognized by pediatric practitioners, who often order extensive and unrewarding testing and physiotherapy treatment.

Methods

Twelve BPT cases observed during a 5-year period (2009–2014) at the Clinical Centre Niš, Niš, Serbia were reviewed. All the subjects were sent to pediatric rehabilitation department by pediatricians for treatment with physiotherapy. Data were collected on the features of torticollis, the age of onset, the duration of episodes, associated symptoms, the frequency of episodes, the persistence of symptoms over time, the age when the disorder finally disappeared, sequelae appearing after the 5th birthday, and family history of BPT, migraine or kinetosis. All the children were followed for periods ranging from 48 to 72 months.

Results and Discussion

The clinical features of included patients are presented in Table 1. The series included 6 females and 6 males.

The features of our cases are generally comparable with those of most cases described in the literature [22–24].

With respect to the age of onset, the frequency and duration of the episodes, associated symptoms and the age of disappearance, our results are similar to other reports [2, 10, 24]. The age at onset of BPT was less than 8 months in 84% of the cases. Episodes of torticollis occurred suddenly on waking in all the cases without any trigger factors. The duration of torticollis ranged from a few hours to a few weeks. In 58% of cases, the condition persisted for more than one week. The frequency of the episodes ranged from once every 3 days to once every 25 days. The head was always turned to the same side in 2 (16.67%) cases, but in 9 (75%) cases torticollis alternated sides with successive attacks.

Accompanying symptoms during attacks of torticollis included irritability (4 cases), vomiting (3 cases), pallor (4 cases) and drowsiness (5 cases). One child presented unstable gait during attacks at the age of 2.5 years. Tortipelvis was observed in 5 (41.67%) cases. The persistence of torticollis during sleep was observed in 2 cases. The episodes were more frequent and lasted longer in the early months and tended to cease as the child became older. The age when episodes ended ranged from 11 months to 62 months. In 11 (91.66%) cases, the disorder disappeared before the patient's 5th birthday. No patient had a family history of BPT. In 10 of the cases, first-degree or second-degree family members had family histories of migraine affecting at least one other family member (4 cases) or ≥ 2 family members (6 cases). In 6 cases, family members had kinetosis. In 5 cases, family members were positive for both migraine and kinetosis.

Table 1

The clinical features of the 12 patients with BPT in infancy

Clinical features	Patients, n (%)
Sex	
female	6/12 (50.00)
male	6/12 (50.00)
Age of onset (months)	
< 3	4/12 (33.33)
3–8	6/12 (50.00)
> 8	2/12 (16.66)
Duration of episodes (days)	
< 1	1/12 (8.33)
1–7	4/12 (33.33)
> 7	7/12 (58.33)
Always on the same side	2/12 (16.67)
Usually on the same side	1/12 (8.33)
Alternation between sides in successive episodes	9/12 (75.00)
Tortipelvis	5/12 (41.66)
Persistence during sleep	2/12 (16.66)
Accompanying symptoms	
irritability	4/12 (33.33)
vomiting	3/12 (25.00)
pallor	4/12 (33.33)
drowsiness/apathy	5/12 (41.67)
Age of disappearance (years)	
< 3	3/12 (25.00)
< 5	8/12 (66.67)
> 5	1/12 (8.33)
Family history of	
migraine	10/12 (83.33)
kinetosis	6/12 (50.00)

BPT – benign paroxysmal torticollis.

Instrumental tests including laboratory tests, brain ultrasound, orthopedic, ophthalmologic and otorhinolaryngologic examinations; ultrasound of the sternocleidomastoid muscle, and EEG were performed in all cases. Each test yielded normal results. The neurological findings were normal in all the cases, both during and after episodes, with no significant changes in muscle tone. A total of 5 (41.67%) children underwent magnetic resonance imaging of the brain. All the results were normal. All the children had normal motor development and normal speech and language development. After the disappearance of BPT, two children developed other forms of periodic syndromes: one boy had migraine with aura, and one girl experienced cyclic vomiting.

Conclusion

Benign paroxysmal torticollis is probably an age-sensitive and migraine-related disorder that is benign in nature. The disorder is often misinterpreted, and children may pointlessly undergo numerous tests. Therefore, it is very im-

portant to recognize and observe this condition in order to avoid extensive, unnecessary and unpleasant procedures on the child. The diagnosis of BPT in infancy is based on clinical grounds: a proper history and physical examination in paticular, and does not call for instrumental tests, which only cause pointless distress to the child and incur unnecessary expense and anxiety on the part of the parents.

The recurrent nature of the episodes in a previously healthy infant is what leads to the diagnosis of BPT.

In cases of torticollis which do not fit into the typical clinical picture of BPT, or if there are any additional symptoms, additional diagnostics is required in order to find the actual cause and avoid making a wrong diagnosis and losing precious time for the treatment of serious diseases.

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