A Rare Case of Spasmus Nutans in an Infant

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ABSTRACT

We report a case of an otherwise healthy 23-month-old boy who presented with nystagmus, head shaking, and abnormal head posture suggestive of spasmus nutans. Neuro-ophthalmologic exam revealed bilateral, low-amplitude, high-frequency, horizontal, disconjugate nystagmus that was more prominent in one eye along with head shaking and a head tilt or face turn. The rest of the exam and the systemic physical examination were normal. Magnetic resonance imaging of the brain did not disclose optic pathway glioma, which has been reported to cause spasmus nutans-like disease. Electroretinogram (ERG) was also recommended to rule out occult retinopathies. However, it was not done due to unavailability of the appropriate corneal electrode for his age. Instead, close follow-up was advised to monitor spontaneous improvement or resolution, or until the child comes of age that he can undergo ERG. This case highlights the management approach and rationale of patients with presumed spasmus nutans. Recognition of the triad of spasmus nutans allows for quick diagnosis and more focused and efficient investigation.

Keywords: pathologic nystagmus, infantile spasms, infant torticollis

INTRODUCTION

Spasmus nutans is a rare, idiopathic, self-limited disorder characterized by the classic triad of nystagmus, head nodding, and anomalous head posture.[1,2] It is generally seen in young children, with an age of onset between 6 to 36 months.[2,3] The characteristic nystagmus in spasmus nutans is sinusoidal, disconjugate, with a frequency of 15 Hz, and an amplitude of up to 2 degrees.[4-6] It has fine and rapid excursions, sometimes described as an "ocular quiver".[7] The head nodding or shaking in spasmus nutans is inconstant, irregular, horizontal, or vertical in direction or both.[3] It generally has a much lower frequency and larger amplitude than the nystagmus and is hypothesized to be a compensatory mechanism rather than a pathologic phenomenon.[4,5] Studies have shown that head-nodding influences eye movements and also disappears during sleep.[5] While the anomalous head position or torticollis accompanying this disease may either be a head tilt or a head turn. In most patients, the disorder spontaneously resolves or abates to subclinical form at 3 to 6 years.[2] However, reported associations with disorders affecting the retina or optic nerves, such as rod-cone dystrophies and optic pathway gliomas, have prompted specialists to regard spasmus nutans as a diagnosis of exclusion.[2] Herein, we describe the clinical presentation, diagnosis, and management of presumed spasmus nutans in a 23-month-old boy.

CASE PRESENTATION

Our patient is a previously healthy, 23-month-old boy who presented with nystagmus of 8-month duration. The

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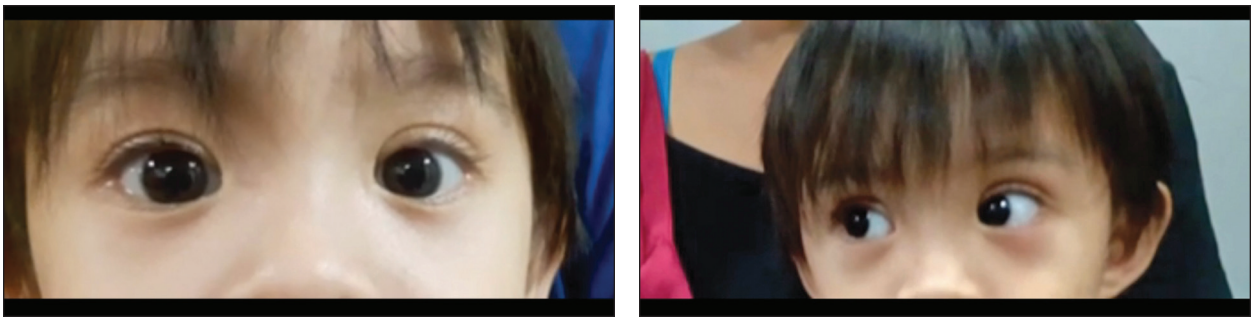

Figure 1. Screenshots of the video show the triad of findings in spasmus nutans. The patient displays a right head tilt and bilateral disconjugate nystagmus that is more prominent on the right eye seen as high-frequency, low-amplitude, "shimmering" horizontal nystagmus. Intermittent head shaking was also observed. When head shaking stops, the nystagmus becomes more perceptible.

patient's mother first noted head shaking (as if signaling "no") which was spontaneous, intermittent, and absent during sleep. The head movement was soon accompanied by bilateral, horizontal, oscillatory eye movements and abnormal head posture. There was no history of head trauma, observations of poor vision, irritability, loss of appetite, fever, seizures, vomiting, and decreased activity.

Past medical and family medical histories were unremarkable. The patient was born full-term in a hospital under an obstetrician. Prenatal, birth, and post-natal periods were likewise unremarkable. He was developmentally at par with peers. Complete immunizations for age were given care of a local health center.

On physical exam, the patient was awake, alert, ambulatory, and not in cardiorespiratory distress. He had normal gross morphology and vital signs. Anthropometric measurements were within acceptable limits for head circumference and a Z score of -2 for height and weight. Systemic and neurologic exams were also unremarkable.

On eye examination, binocular visual acuity was 20/130 using the Teller II Acuity Cards, typical for age.[8] He demonstrated no eye preference. Cycloplegic refraction was +2.0 diopter sphere in each eye which was non-amblyogenic for age. Pupils were 3 millimeters in size, equally and briskly reactive to light, and with no relative afferent pupillary defect (RAPD). The eyes were orthophoric on primary gaze by Hirschberg light reflex with full ocular motility. On close examination, the patient displayed a "shimmering," pendular, low-amplitude, high-frequency, horizontal nystagmus on both eyes along with horizontal head shaking and head tilt. At times, the nystagmus appeared monocular (Figure 1). Head immobilization worsened the nystagmus. The nystagmus and head shaking were accompanied with anomalous head posture, either a head tilt or a face turn in variable direction. The anterior segment of both eyes was unremarkable. Fundoscopy showed normal-looking optic discs, macula, and peripheral retina.

A magnetic resonance imaging (MRI) of the brain was done, which did not disclose any abnormality of the afferent visual pathway; specifically, it was negative for optic pathway glioma. Ideally, an electroretinogram (ERG) is part of the workup of spasmus nutans[9]; however, the test could not be carried out due to unavailability of the appropriate corneal electrode for his age.

A repeat eye examination six months after the initial visit disclosed persistence of the triad of nystagmus, head shaking, and head tilt. Repeat refraction was done, which was non-amblyogenic for age. The patient remained healthy and had not developed any new neurologic signs and symptoms.

DISCUSSION

Salient features of the case include sudden onset of abnormal head and eye movements and anomalous head posture in a 23-month-old boy. Ocular examination showed bilateral, disconjugate, pendular, low-amplitude, high-frequency nystagmus that is more prominent in one eye associated with horizontal head shaking and anomalous head posture, either a head tilt or a face turn. Anatomically, the afferent visual system was normal. MRI of the brain did not show any optic pathway abnormalities. A diagnosis of presumptive spasmus nutans was made based on the presence of disconjugate nystagmus, head shaking and abnormal head tilt in an infant.

In suspected cases of spasmus nutans, a thorough work-up is important as there are reports of patients with spasmus nutans-like nystagmus with underlying ocular, intracranial, or systemic abnormalities on investigation.[2,10-12] In a chart review of 22 patients diagnosed with spasmus nutans, only three had no associated ocular, intracranial, or systemic conditions and can be truly labeled idiopathic. Most patients had a significant refractive error, strabismus, chiasmal gliomas, or rod/cone dystrophy.[2] Of note, optic pathway glioma, a sight-threatening but potentially treatable disease, should be excluded in patients who present spasmus nutans-like disease.[10,13] Its true incidence is low among children with spasmus nutans with normal neurologic function and in the absence of a RAPD or optic disc pallor,[3,14,15] but a

published case report of spontaneously resolving spasmus nutans in an 8-month-old infant with chiasmal glioma serves as a cautionary tale.[13] Thus, patients with spasmus nutans must undergo further investigations to exclude other potentially treatable causes. Recommended minimum diagnostic workup includes a brain MRI and ERG.[2,9,16,17]

A full-field ERG, while recommended, was not performed due to the unavailability of the appropriate corneal electrode for the patient's age. ERG is most beneficial to diagnose or rule out occult retinopathies that have reported associations with spasmus nutans, like rode/cone dystrophies, achromatopsia, and congenital stationary night blindness.[9,11,18] In place of this test, we stressed the importance of close follow-up with the patient's mother. The diagnosis of spasmus nutans remains presumptive until we see a spontaneous resolution through time or the patient comes to an age that he can undergo an ERG test.

Meanwhile, monitoring of visual acuity, refraction, and ocular alignment to check for amblyopia and strabismus are also crucial in the follow-up of young patients with spasmus nutans. In a study by Young et al., out of 18 pediatric patients with spasmus nutans, 8 had clinical amblyopia requiring occlusion therapy, and 11 patients had improved visual acuity with spectacle correction.[6] They concluded that early intervention and treatment of anticipated visual problems might optimize visual outcomes in patients with spasmus nutans. Thus, we advocate the same principles and continue to monitor vision, refraction, and ocular alignment in this patient.

CONCLUSION

In conclusion, this case demonstrates the management approach to a young patient presenting with nystagmus, head nodding, and abnormal head posture. While the diagnosis of spasmus nutans is made clinically based on the appreciation of a triad of findings, a complete neuro-ophthalmologic exam and ancillary tests (specifically brain imaging and ERG) are required to rule out any underlying cause. Lastly, monitoring for significant refractive error, amblyopia, and strabismus should be carried out on all patients with spasmus nutans.

Ethical Consideration

A signed informed consent for data collection, video recording, and case presentation was provided by the patient's mother.

Statement of Authorship

Both authors contributed in the conceptualization of work, acquisition and analysis of data, drafting and revising of manuscript, and final approval of the version to be published.

Author Disclosure

Both authors declared no conflicts of interest.

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