

Spasmus Nutans and Congenital Ocular Motor Apraxia With Cerebellar Vermian Hypoplasia

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Background: Spasmus nutans and congenital ocular motor apraxia share clinical characteristics. However, their development in a patient with cerebellar vermian hypoplasia has not been previously described.

Objective: To report spasmus nutans and congenital ocular motor apraxia in a child with cerebellar vermian hypoplasia.

Design: Case report.

Setting: Tertiary-care hospital.

Patient: A 7-year-old boy with a history of spasmus nutans during infancy and developmental delay was referred for the evaluation of abnormal head and eye movements.

Results: The patient had impaired voluntary saccades and smooth pursuit in the horizontal plane and showed thrusting movements of the head during attempted gaze shift. Magnetic resonance imaging of the brain demonstrated cerebellar vermian hypoplasia, especially in the inferior portion.

Conclusions: Spasmus nutans and congenital ocular motor apraxia may develop in patients with cerebellar vermian hypoplasia. In patients with congenital ocular motor apraxia, a history of spasmus nutans should be sought, and careful evaluation of the cerebellar vermis is needed during brain imaging.

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SPASMUS NUTANS refers to a syndrome of nystagmus, head nodding, and abnormal head position.¹ The nystagmus is intermittent and consists of fine, pendular, and predominantly horizontal oscillations of high frequency (up to 15 Hz) and a variable-phase relationship between the eyes.^{2,3} The head nodding is also intermittent and is usually observed with the nystagmus. Congenital ocular motor apraxia is characterized by impaired voluntary saccades and abnormal head thrusts.⁴ Since these patients cannot generate a saccade, they turn their heads quickly in the direction of the visual target, generating the characteristic head thrust. Spasmus nutans and congenital ocular motor apraxia develop during early childhood and have a self-limited course.

The neural substrates of spasmus nutans and congenital ocular motor apraxia remain unidentified. These conditions do not normally accompany other neurological abnormalities. However, various intracranial lesions may be manifested as symptoms of spasmus nutans⁵ or ocular motor apraxia.⁶ We herein report a case

of congenital ocular motor apraxia in a child in whom spasmus nutans developed in the neonatal period. The only abnormal finding in the magnetic resonance imaging of the brain was cerebellar vermian hypoplasia. To our knowledge, this is the first report of spasmus nutans and congenital ocular motor apraxia in a patient with cerebellar vermian hypoplasia.

REPORT OF A CASE

A 7-year-old boy was referred by a pediatrician for the evaluation of impaired eye movements and head thrusting during attempted gaze shift. The patient had an uneventful prenatal period and no family history of ophthalmological or neurological diseases. During the neonatal period, he was admitted to our hospital with a diagnosis of sepsis secondary to pneumonia. At that time, he was found to oscillate his head. The movement was found to be associated with horizontal oscillations of the eyes. He also had esotropia. The abnormal head and eye motions were mainly induced by external stimuli and lasted approximately 30 seconds, subsiding during

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Figure 1. Head thrust during gaze shift. On attempted gaze shift with the head free, the patient thrusts his head toward the object of attention (A and B). This head thrust is characteristically accompanied by a contraversive deviation of the eyes, necessitating an overshoot of the head movement for fixation (B). Subsequently, the head slowly returns to a position that allows the eyes to adopt the primary position while observing the target (C).

sleep. He was diagnosed as having spasmus nutans. During the first few months of life, he was thought to be blind, because he was unable to follow objects with his eyes and had difficulty initiating eye contact. However, ophthalmological examinations, including visual evoked potentials, showed no abnormal findings. His developmental milestones were also delayed. He could sit unsupported at 8 months and stand without assistance at 18 months of age. He was able to take steps at 24 months of age. However, his mental development was within the reference range for his age. The spasmus nutans had resolved by 2 years of age with some fluctuation, but at about that time, his parents noticed intermittent head thrusting, especially during attempted gaze shift, which persisted until the consultation.

The general physical and ophthalmological examination findings were normal, and the neurological examination showed a joyful child with normal mentation for his age. However, when he was asked to fixate an object on either side with the head free, he thrust his head toward the object of attention. This was characteristically accompanied by a contraversive deviation of the eyes, necessitating an overshoot of the head movement to achieve fixation (**Figure 1**). Moreover, with the head fixed, he could not make any horizontal saccades in the desired direction. However, he showed intermittent reflexive saccades in the horizontal direction. Horizontal smooth pursuit was also impaired. Sometimes blinking preceded attempted horizontal saccades and smooth pursuit. The vertical saccades and smooth pursuit were preserved. Using the passive head rotation, the vestibulo-ocular reflex was determined to be normal. Other findings of the neurological examination were normal. Magnetic resonance imaging of the brain demonstrated cerebellar vermian hypoplasia, mainly in the inferior portion (**Figure 2**).

COMMENT

Spasmus nutans developed in our patient with congenital ocular motor apraxia in the neonatal period. Spasmus nutans and congenital ocular motor apraxia share the characteristics of abnormal head and eye motion (predominantly involving the horizontal plane), the recruitment of head motion to compensate for abnormal eye motion, a self-limiting course, and familial occurrence. These disorders usually develop in infancy. Spasmus nutans

mostly resolves spontaneously within 1 to 2 years after its appearance, although subtle abnormalities may persist in eye movement recordings.⁷ In cases of congenital ocular motor apraxia, the typical head-thrusting behavior emerges usually around 4 months of age, when adequate head control is achieved. Congenital ocular motor apraxia also tends to improve with age. Both syndromes are occasionally familial and have been reported in monozygotic twins.^{8,9}

In a recording of spasmus nutans, the nystagmus consisted of sinusoidal and disjunctive eye movements at a frequency of 11 Hz and an amplitude of 2°. Such ocular oscillations may be conjugate, disjunctive, or purely monocular.³ The nystagmus in spasmus nutans has a high-frequency component, which is similar to the principal frequency components of saccadic eye movements and suggestive of an imbalance of the saccadic pulse-generating mechanism.² The head nodding was recorded to consist of a 3-Hz sinusoidal horizontal oscillation with an amplitude of 3°. It abolishes or dampens the nystagmus and permits the discrimination of the finer features of the visual stimuli.^{2,10} Measurements of eye movements in congenital ocular motor apraxia documented delayed and hypometric voluntary saccades with normal velocity.¹¹ The head thrusts in congenital ocular motor apraxia are also adaptive behavioral patterns. During head thrusts, the eyes are brought to a position of extreme contraversive deviation with the intact action of the vestibulo-ocular reflex. As the head continues to move past the target, the eyes are dragged along in space until they become aligned with the target. Then the head rotates backward to bring the eyes to the primary position while maintaining a fixation. In older patients, head thrusts may be used to boost the saccadic command. Sometimes, blinking precedes the intended eye movements. Pause cells stop discharging during blinks,¹² and stimulation of the pause cell in monkeys inhibits blinking.¹³ This link between saccades and blinking via the pontine pause cells may be responsible for the compensatory synkinetic blink to shift direction of gaze in congenital ocular motor apraxia.¹⁴ The pathomechanisms of spasmus nutans and congenital ocular motor apraxia have not been elucidated. Both disorders are primarily idiopathic and are not accompanied by other neurological or extraneurological abnormalities, although strabismus, developmental delay, and clumsiness may be associated.^{1,4} However, a recent study showed that vermian hypopla-

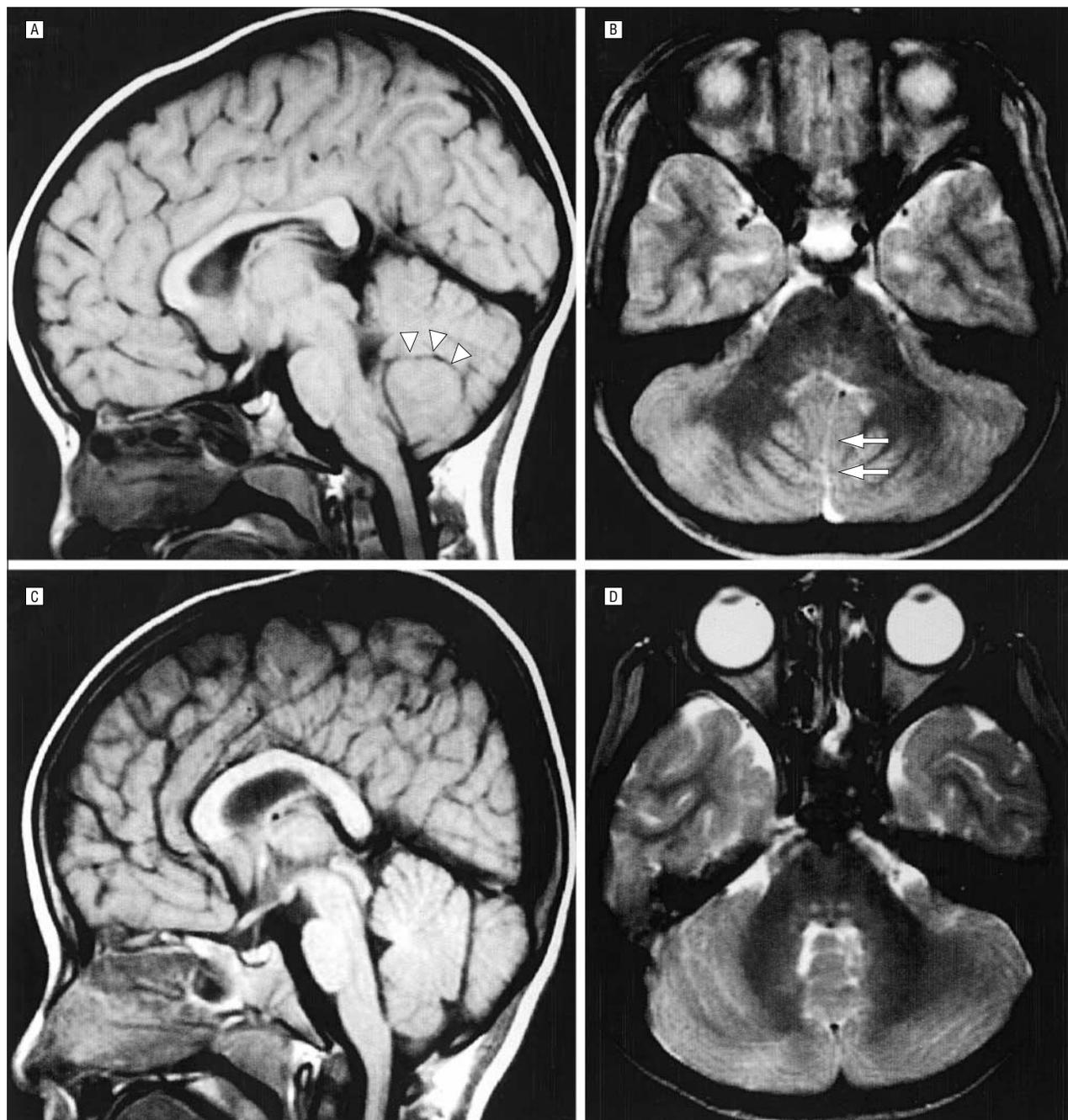


Figure 2. Cerebellar vermal hypoplasia. Magnetic resonance imaging of the brain demonstrates vermal hypoplasia, especially in the inferior portion (arrowheads) (A). The nodulus is absent and the cerebellar tonsils are closely apposed (arrows) (B), compared with the findings in a healthy control subject (C and D).

sia, which preferentially involves the inferior portion, is the most common finding in children with congenital ocular motor apraxia.¹⁵ Our patient also showed hypoplasia of the inferior vermis. The posterior vermis may be involved in saccade control.¹⁶ Abnormal saccades may develop as a result of posterior vermal infarction.¹⁷ Ablation of the vermal lobules VI and VII in primates has been shown to induce hypometric and delayed saccades,¹⁸ and abnormalities in the cerebellar vermis are closely correlated with the eye movement deficits that characterize congenital ocular motor apraxia.¹⁹ Some of the vermal Purkinje cells discharge 11 to 24 milliseconds before the

onset of saccades.²⁰ Moreover, microstimulation of an area within lobule VII of the vermal cortex evokes saccades, and lesions of this area abolish these saccades.²¹ Vermal lobules IV through VII receive mossy fiber inputs from the paramedian pontine reticular formation, identified as a premotor area for the horizontal saccades.²² Purkinje cells in the vermis normally inhibit activity in the fastigial nucleus, and the caudal fastigial nucleus projects bilaterally to the rostral poles of the superior colliculus,²³ where the fixation cells reside.²⁴ Hypoplasia of the vermal cortex could disinhibit the caudal fastigial nucleus, which leads to an abnormally increased activity in the

fixation region of the superior colliculus and results in the inhibition of saccadic generation. The pathophysiological role of cerebellar vermian hypoplasia in the generation of spasmus nutans remains unclear. However, the shared characteristics between congenital ocular motor apraxia and spasmus nutans suggest a common pathophysiology rather than a mere coincidence.

CONCLUSIONS

We described a child with congenital ocular motor apraxia and spasmus nutans associated with cerebellar vermian hypoplasia. In patients with congenital ocular motor apraxia, a history of spasmus nutans should be sought, and careful evaluation of the cerebellar vermis is needed during the brain imaging.

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