CLINICAL NOTE

Congenital Torticollis and Saccular Dysfunction

A Case Report

Ashley Hallberg, AuD; Robert T. Standring, MD; Syed Ahsan, MD

Importance: This is the first report, to our knowledge, of a child with torticollis due to saccular dysfunction.

Observation: An 18-month-old infant with torticollis was referred for postural imbalance and observed rotary nystagmus. The infant had undergone physical therapy treatment of left torticollis for nearly 15 months. Cervical vestibular evoked myogenic potentials (cVEMPs) were recorded to assess saccular function and caloric stimulation and positional and rotational testing were performed to evaluate other vestibular receptors. The child demonstrated abnormal cVEMP findings, with a low-amplitude response on the left, which indicated left-sided saccular dysfunction. The patient's rotary-torsional nystagmus suggested positional vertigo secondary to abnormal saccular function.

Conclusions and Relevance: This case highlights that saccular dysfunction should be considered when timely resolution of congenital torticollis is not obtained with physical therapy. Early detection of abnormal saccular function in infants and young children with CPT is necessary to ensure appropriate intervention. Further study needs to be done to confirm our findings.

JAMA Otolaryngol Head Neck Surg. 2013;139(6):639-642

TORTICOLLIS IS A TERM USED to describe a shortened or fibrotic sternocleidomastoid muscle (SCM), which can lead to tilting of the head toward the tightened side. Usually presenting at birth or soon after, congenital torticollis has an estimated incidence of 1 in 250 live births.[1] Classically, the etiology of torticollis has been hypothesized to arise from congenital muscular abnormality; abnormal postural position; and osseous, neurological, or ocular abnormalities. Pediatric evaluation of suspected congenital torticollis should be comprehensive and include a complete physical examination, neurological assessment, plain radiographs, and ophthalmological examination.

The exact cause of congenital muscle torticollis (CMT) remains unknown. Research suggests that SCM injury causes fibrotic tissue formation, which results in shortening of the muscle. Signs of CMT include unilateral fibrosis or shortening of the SCM, lateral flexion of the head, and deviation of the chin to the contralateral side. Congenital muscle torticollis produces loss of active and passive range of motion and generally a mass is palpable in the SCM during the first 3 months of life. Compensatory changes can also occur in posture, and children can develop flattening of parietal-occipital and often frontal areas from preferential sleeping posture.[2,3]

In contrast, congenital postural torticollis (CPT) has signs similar to those of CMT but with no palpable mass in the SCM. Patients with CPT tend to have decreased ability to actively rotate or laterally flex their head but have normal passive cervical range of motion. They tend to intermittently tilt their head rather than stay fixed, as in CMT. Congenital postural torticollis may be due to deformational plagiocephaly at birth or 1-sided position after birth.[2,4]

Osseous abnormalities, such as Grisel syndrome (C1-C2 subluxation), may also produce torticollis. This often arises from an infectious etiology, such as upper respiratory tract infection, otitis media, sinusitis, pharyngitis, cervical adenitis, or retropharyngeal abscess.[1,5] Neurological abnormalities, such as posterior fossa tumors, syringomyelia, and spinal cord tumors, can lead to torticollis. Neurological causes are differentiated by the presence of other symptoms, such as headaches, nausea, vomiting, and positive neurological signs.[1,6] Ocular abnormalities may also cause torticollis. Infants with extraocular

Author Affiliations:
Department of Otolaryngology–Head and Neck Surgery, Henry Ford Hospital, Detroit, Michigan.

Figure 1. An 18-month-old infant demonstrating left lateral tilt and rotation of the head to the right side.

impairment in 1 eye adapt an abnormal posture, a tilt of the head, which may help with binocular vision and avoid diplopia. Paresis of the superior oblique muscle is the most common source of ocular torticollis. Symptoms generally occur later than nonocular torticollis, after the infant has developed head control and binocular vision. There may be associated nystagmus with restrictive ocular movement but no cervical range of motion restrictions.[7]

We present a unique case, the first report to our knowledge, of a child with CPT due to abnormal saccular function.

REPORT OF A CASE

An 18-month-old male infant with a history of CPT was referred to us for evaluation of possible imbalance issues. He demonstrated left lateral tilt and rotation of the head to the right side (**Figure 1**) but was noted to have a full range of motion, and his left lateral tilt was intermittent. Findings from pediatric ophthalmologic and neurologic examinations were unremarkable, and results from neck ultrasonography to rule out muscle fibrosis were normal.

At 2 months of age, the infant presented to the physical therapy (PT) department with a classic torticollis posture. A differential diagnostic workup was completed to determine the cause and institute appropriate intervention. Physical therapy treatment consisted of positioning, strengthening, stretching, and neuromuscular facilitation techniques to assist the infant to develop gross motor skills, appropriate balance, and a midline head position. A home stretching and exercise program was also incorporated. After 15 months of PT and home stretching exercises, the infant demonstrated some gain of lateral flex of the head but continued to exhibit imbalance difficulties, primarily with postural stability. Difficulty maintaining the head in midline also persisted. On later PT observation, spontaneous rotational nystagmus was noted for approximately 10 to 15 seconds in duration. This resolved with head movement from lateral tilt to midline orientation as well as ocular tracking.

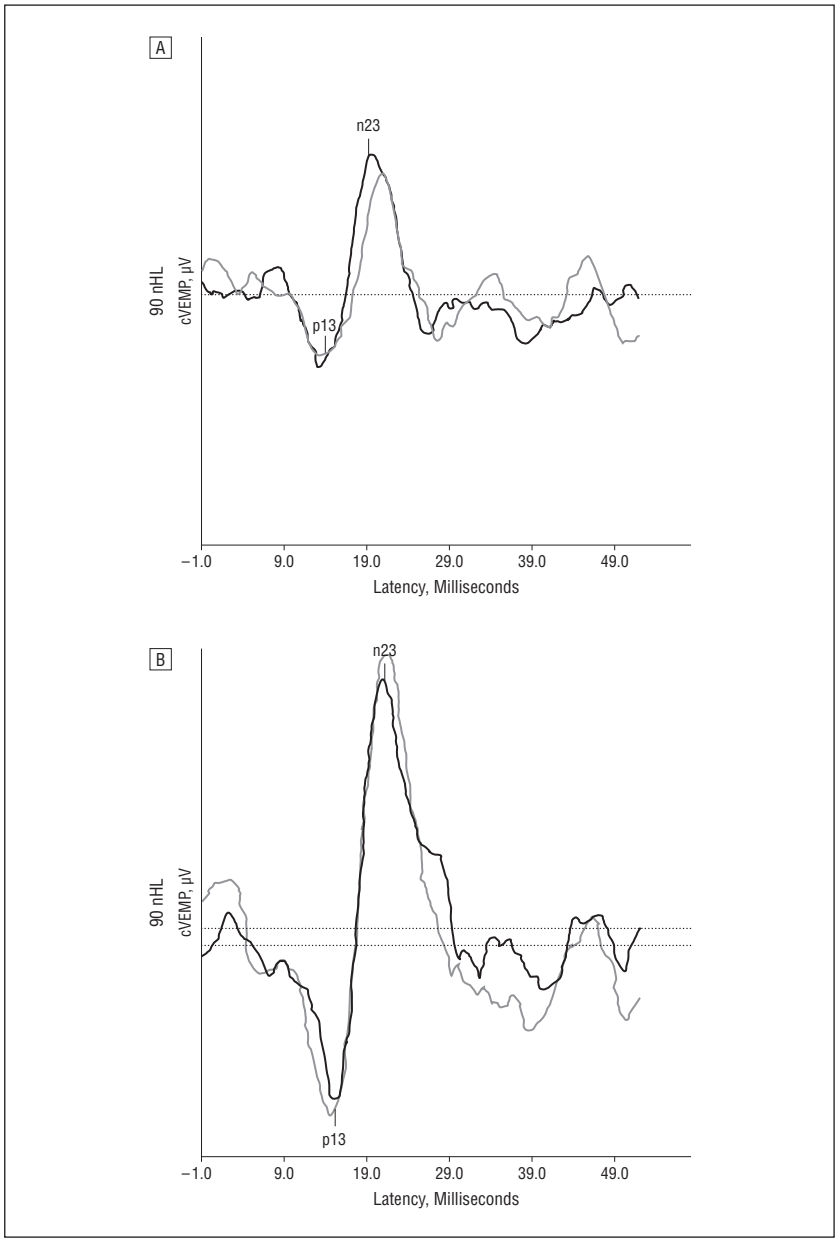

Figure 2. Asymmetric cervical vestibular evoked myogenic potential (cVEMP) testing generated in response to air conduction 500 Hz tone bursts was reduced in amplitude by 69% on the left (affected) side (A) compared with responses on the right (normal) side (B), indicating left-sided saccular dysfunction (normal symmetry is <40%). p13 and n23 indicate the p13-n23 complex.

Our physical examination revealed obvious postural instability, tightness of the left SCM, and a less prominent right SCM. The infant otherwise had normal development and had reached age-appropriate milestones. Findings from an audiologic evaluation were unremarkable. Cervical vestibular evoked myogenic potential (cVEMP) testing was performed to evaluate sac-

cular function. Abnormal, asymmetric cVEMP amplitudes of the p13-n23 complex were recorded for the left (affected) and the right (normal) sides. A 69% amplitude reduction on the left side (**Figure 2**) indicated saccular dysfunction or disruption within the saccule pathway (inferior vestibular nerve involvement). There was no pattern of hypersensitivity to sound reflected by cVEMP findings. Caloric testing was symmetrical, and rotational chair sinusoidal harmonic acceleration study revealed a normal pattern. Rotary-torsional nystagmus on right positional testing suggested a presence of positional vertigo secondary to abnormal saccular function.

The patient underwent 1 year of vestibular physical therapy focusing on saccular function and retraining. Twelve months after his original neurotologic workup, he underwent repeated cVEMP testing, which demonstrated a normal test. The vestibular evoked myogenic potential (VEMP) responses were present bilaterally with appropriate latencies, with only a 20% amplitude reduction on the left side compared with the previous 69% reduction. There was no pattern of hypersensitivity to sound stimulation (**Figure 3**).

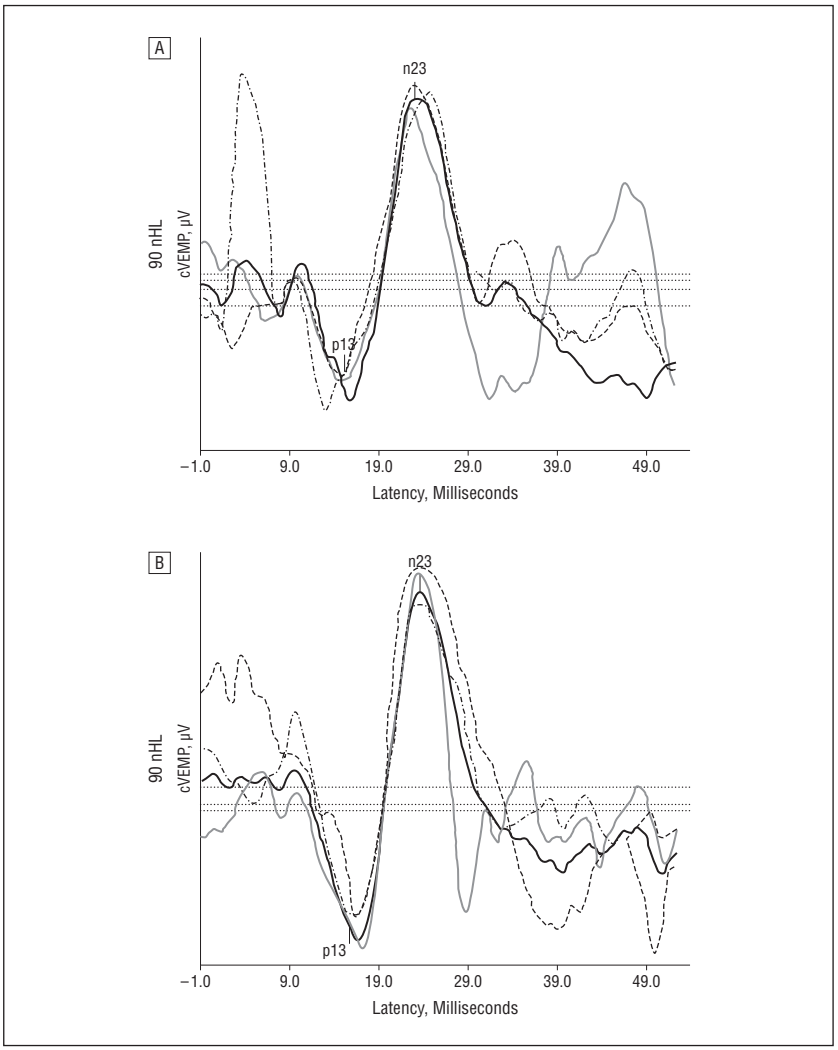

Figure 3. Repeated cervical vestibular evoked myogenic potential (cVEMP) testing after 1 year of vestibular rehabilitation demonstrated minimally asymmetric reduced amplitude (20%) on the left (affected) side, confirming improvement of saccular function. The right side is normal. p13 and n23 indicate the p13-n23 complex.

DISCUSSION

Peripheral vestibular loss in young children commonly presents with delayed motor development and postural abnormalities, including head tilt toward the side of lesion and disequilibrium.[8,9] Additional manifestation of unilateral vestibular loss is static torsion of the eyes toward the side of the lesion. This ocular tilt reaction is likely caused by unilateral loss of otolithic function.[10] The role of the otolith organs, the maculae of the utricle and saccule, is to sense linear accelerations in the horizontal and vertical planes as well as to sense static orientation of the head by responding to dynamic changes in linear velocity. Disruption to the otolith organs, or more specifically the sacculocollic pathways, can influence the development of postural control and put children at risk for impairment of sensory information critical to normal development of locomotion and motor coordination.[11,12]

To our knowledge, an underlying saccular dysfunction has not been previously considered to be part of the differential diagnosis for CPT. Early diagnosis and determination of the cause of torticollis are imperative to recommend further workup and develop the most efficacious treatment plan. As an example, PT is most effective for CPT when performed at 2 to 8 months of age.[4] Some patients, however, do not respond to PT and would then benefit from further testing for saccular dysfunction as the cause of their head tilt.

Experimental animal model studies have found the early postnatal period to be a critical time for the vestibular system to provide input to neuromuscular development of postural muscles and the development of balance strategies.[13,14] Saccular function can be noninvasively assessed by use of cVEMPs. Although not commonly used in infants, cVEMPs offer an objective test to explore the vestibular system and the sacculocollic pathways in infants and very young children.[11] The use of cVEMP testing can refine the differential diagnosis of CPT to avoid delayed motor development and postural impairment due to saccular deficits.

In our patient, abnormal cVEMP responses were observed on the left side, indicating that the left lateral head tilt was attributed to saccular dysfunction and resultant CPT. The mechanism of the effects of saccular dysfunction on neck position may be due to abnormal sacculocollic reflex as well as abnormal input from

the saccule to the otolith-ocular system along with the utricle.[15] In addition, alteration of the vestibulospinal subsystem may contribute.

It should be noted that performing vestibular tests on infants and children requires a significant effort with respect to time and modification of the routine testing conditions. One of the intrinsic problems with doing VEMP testing in very young children, who often cannot cooperate and/or condition to contracting the SCM independently, is assuring neck muscle activation and maintaining consistent contraction. The tonic state of the SCM muscle is vital in the recording activity. Children (typically those ≥4 years) can be laid supine on an examination table and asked to raise their head off of the table to activate or contract the neck flexors. Infants, however, are not cooperative to this task, limiting the ability to obtain VEMPs. To activate muscle activity and maintain consistent SCM contraction between test sides, a restraint method was used with our patient. Parental consent was obtained. The patient was wrapped in a blanket restraint and then set on a parent's lap for additional restraint and parental comforting. The patient's head was then turned and held by an assistant while the electromyography (EMG) was monitored by the audiologist to maintain consistent amplitude of EMG (between the right and left sides) during recordings. Without the use of an EMG monitor, having an assistant hold the patient's head could contribute to false or abnormal findings. But we used an EMG monitor to record contraction levels of the sternocleidomastoid muscle to ensure the same level of contraction.

In conclusion, the findings of the present case suggest that saccular dysfunction plays a possible role in infants with congenital postural torticollis. Early detection of abnormal saccular function in infants and young children with CPT is necessary to ensure appropriate intervention and prevent development of impaired motor coordination, reduced locomotion, and postural instability. The exploration of potential saccular deficit(s) in pediatric patients with CPT who fail to respond to PT, especially those demonstrating imbalance or lack of postural control, is warranted. Larger studies may be undertaken to validate these findings.

Submitted for Publication: November 30, 2012; final revision received January 13, 2013; accepted February 5, 2013.

Correspondence: Syed Ahsan, MD, Department of Otolaryngology–Head and Neck Surgery, Henry Ford Hospital, 2799 W Grand Blvd, Detroit, MI 48202 (sahsan3@hfhs.org).

Author Contributions: All authors had full access to all the data in the study and take responsibility for the integrity of the data and the accuracy of the data analysis. *Study concept and design:* Ahsan. *Acquisition of data:* Hallberg and Ahsan. *Analysis and interpretation of data:* All authors. *Drafting of the manuscript:* Hallberg and Ahsan. *Critical revision of the manuscript for important intellectual content:* All authors. *Administrative, technical, and material support:* Hallberg and Standring. *Study supervision:* Ahsan.

Conflict of Interest Disclosures: None reported.

Previous Presentation: This study was a poster presentation at Triologic Combined Sections Meeting; January 26-28, 2012; Miami Beach, Florida.

REFERENCES

1. Luther BL. Congenital muscular torticollis. *Orthop Nurs*. 2002;21(3):21-27.
2. Gray GM, Tasso KH. Differential diagnosis of torticollis: a case report. *Pediatr Phys Ther*. 2009; 21(4):369-374.
3. Cheng JC, Tang SP, Chen TM, Wong MW, Wong EM. The clinical presentation and outcome of treatment of congenital muscular torticollis in infants: a study of 1,086 cases. *J Pediatr Surg*. 2000; 35(7):1091-1096.
4. van Vlimmeren LA, Helders PJ, van Adrichem LN, Engelbert RH. Torticollis and plagiocephaly in infancy: therapeutic strategies. *Pediatr Rehabil*. 2006; 9(1):40-46.
5. Berry DS, Moriarty RA. Atlantoaxial subluxation related to pharyngitis: Grisel's syndrome. *Clin Pediatr (Phila)*. 1999;38(11):673-675.
6. Parikh SN, Crawford AH, Choudhury S. Magnetic resonance imaging in the evaluation of infantile torticollis. *Orthopedics*. 2004;27(5):509-515.
7. Williams CR, O'Flynn E, Clarke NM, Morris RJ. Torticollis secondary to ocular pathology. *J Bone Joint Surg Br*. 1996;78(4):620-624.
8. Brookhouser PE, Worthington DW, Kelly WJ. Unilateral hearing loss in children. *Laryngoscope*. 1991;101(12, pt 1):1264-1272.
9. Halmagyi GM, Curthoys IS. Clinical changes in vestibular function over time after lesions: the consequences of unilateral vestibular deafferentation. In: Herdman SJ, ed. *Vestibular Rehabilitation*. Philadelphia, PA: F.A. Davis; 1994:90-108.
10. Barin K, Durrant J. Applied physiology of the vestibular system. In: Canalis RF, Lambert PR, eds. *The Ear: Comprehensive Otology*. Philadelphia, PA: Lippincott Williams & Wilkins; 2000:113-140.
11. Sheykholeslami K, Megerian CA, Arnold JE, Kaga K. Vestibular-evoked myogenic potentials in infancy and early childhood. *Laryngoscope*. 2005; 115(8):1440-1444.
12. Shall MS. The importance of saccular function to motor development in children with hearing impairments. *Int J Otolaryngol*. 2009;2009:972565.
13. Van Cleave S, Shall MS. A critical period for the impact of vestibular sensation on ferret motor development. *J Vestib Res*. 2006;16(4-5):179-186.
14. Christensson M, Garwicz M. Time course of postnatal motor development in ferrets: ontogenetic and comparative perspectives. *Behav Brain Res*. 2005;158(2):231-242.
15. Murofushi T, Ushio M, Takai Y, Iwasaki S, Sugasawa K. Does acute dysfunction of the saccular afferents affect the subjective visual horizontal in patients with vestibular neurolabyrinthitis? *Acta Otolaryngol Suppl*. 2007;(559):61-64.