Published in final edited form as:

Ophthalmol Plast Reconstr Surg. 2012 March ; 28(2): e36–e39. doi:10.1097/IOP.0b013e3182191be3.

Non-Ophthalmic Symptoms Secondary to Ocular Torticollis From Severe Blepharoptosis: An Underappreciated But Treatable Condition

Brenda L. Bohnsack, M.D., Ph.D.¹, Rina Bhatt, MS (India), F.R.C.S(I), F.R.C.S(Ed)¹, and Alon Kahana, M.D., Ph.D.^{1,2}

¹Department of Ophthalmology and Visual Sciences, Kellogg Eye Center, University of Michigan, Ann Arbor, Michigan

Abstract

Purpose—Identify systemic symptoms associated with ocular torticollis due to bilateral blepharoptosis.

Methods—Retrospective chart review identified patients with ocular torticollis due to bilateral congenital or acquired blepharoptosis who underwent surgical correction (levator resection or frontalis sling). Pre-operative and post-operative assessment of systemic symptoms in adults and developmental milestones are reported.

Results—We present 4 cases, one adult and three young children, with severe bilateral upper eyelid ptosis. In the adult patient, it caused debilitating back pain that required orthopedic evaluation, but was completely relieved with ptosis repair surgery. In the toddlers, subjective developmental motor delay was rapidly reversed following ptosis repair surgery.

Conclusions—We suggest that primary care physicians, as well as spine and neuromuscular specialists, consider the possibility of ocular torticollis due to eyelid ptosis in the evaluation of patients with unexplained back and/or neck pain, or with gross motor developmental delay. We propose that symptomatic ocular torticollis is an indication for ptosis repair.

Introduction

Ocular torticollis refers to abnormal head position secondary to ophthalmic conditions, driven by a conscious or unconscious effort to maintain a clear visual axis and preserve binocularity¹. Reported ophthalmic causes include strabismus (e.g. head tilt associated with superior oblique underaction), and nystagmus (with a null-point outside of primary gaze)¹. Symptoms associated with torticollis are numerous, including severe back and neck pain, muscle spasms, and even facial asymmetry. We now expand on the role of severe bilateral upper eyelid ptosis, acquired or congenital, as a cause of ocular torticollis with systemic symptoms that can be corrected by surgically addressing the ptosis (figure 1A).

We present four cases of severe bilateral blepharoptosis causing systemic symptoms due to ocular torticollis. Preoperatively, the adult patient reported severe back and neck pain for which he had sought orthopedic evaluation. Two of the infants were reported by each set of parents to have gross motor developmental delay. Following surgical correction of the blepharoptosis, the patients regained normal head position. Furthermore, the adult patient reported resolution of his neck and back pain and the infants quickly attained the appropriate

²Corresponding Author: 1000 Wall Street, Ann Arbor, MI 48105, Telephone Number: 734-936-8654, Fax Number: 734-615-0542, akahana@med.umich.edu.

developmental milestones. We suggest that ocular torticollis due to eyelid ptosis is an important diagnosis to be considered by primary care physicians in patients with unexplained back and/or neck pain, or with gross motor developmental delay. We propose that early surgical intervention be considered in the presence of symptomatic ocular torticollis secondary to ptosis.

Methods

A retrospective chart review was performed on four patients with congenital or acquired severe bilateral blepharoptosis who presented to the Eye Plastic and Orbital Surgery Clinic at the University of Michigan Kellogg Eye Center. Preoperative examination included complete history, ophthalmic examination, and external photographs. Each patient underwent ptosis repair with bilateral placement of silicone frontalis slings. Bilateral lateral canthoplasty was also performed on patient 3. Postoperative examination included external photographs and subjective assessment of systemic symptoms. Informed consent for use of photographs was obtained from each of the patients and the study was approved by the University of Michigan Institutional Review Board.

Results

Case 1

A 62-year-old man presented to his primary care physician with a 3-year history of progressive upper back pain localized to the upper thoracic vertebrae and lower neck. The patient was an avid triathlete, bicycling 50–100 miles weekly, but was experiencing increased difficulty in training due to back pain. The primary care physician referred him to a spine specialist, who found modest osteopenia and recommended physical therapy. The pain persisted despite physical therapy, and surgical intervention was considered.

Concurrently, the patient developed a progressive dysphagia that was associated with upper chest pain, a lump in his throat and general pharyngeal tightness. Comprehensive blood tests were all within the normal range. In addition, testing for *Helicobacter* was negative. The patient underwent an upper endoscopy that revealed mild esophagitis, and a proton-pump inhibitor was prescribed.

At the same time of these evaluations, the patient presented to the Eye Plastics, Orbital, and Facial Cosmetic Surgery Clinic at the Kellogg Eye Center for evaluation of bilateral blepharoptosis. He reported progressive bilateral upper lid drooping for the past decade. His past ophthalmic history included a negative workup for myasthenia gravis, and bilateral levator resection surgery 5 years earlier that addressed the ptosis for only a few months.

Upon further questioning, the patient reported that his mother, maternal aunt and maternal grandfather all suffered from severe bilateral upper lid blepharoptosis. He denied any French-Canadian heritage. His mother started having difficulty with the blepharoptosis in her 50s and had undergone multiple eyelid surgeries to correct blepharoptosis, eventually suffering from severe and very debilitating dry eyes. She also required gastric-tube feeding toward the end of her life.

On examination, the patient's visual acuity was 20/30 on the right and 20/40 on the left. His extraocular motility revealed mild limitation of supraduction, but he denied diplopia in any gaze. External examination showed severe bilateral blepharoptosis with the upper lid margins positioned below the corneal reflexes in both eyes (figure 1B). In addition, he had poor upper lid excursion (figure 1C). Due to his blepharoptosis, the patient unconsciously extended his neck and maintained head retroflexion with a chin-up position. In addition, he

had strong frontalis flexion. Tangent visual field testing revealed superior fields of 2 degrees on the right and 0 degrees on the left which improved to 30 degrees bilaterally with lid elevation. Schirmer's tear testing was normal bilaterally and he had no evidence of dry eye syndrome. The remainder of his examination was unremarkable.

Based on his evaluation, a clinical diagnosis of oculopharyngeal muscular dystrophy (OPMD) was made, and ptosis repair with silicone frontalis slings was recommended². Genetic testing for the PABP2 gene demonstrated 10 GCG repeats in allele 1 and 6 repeats in allele 2 (normal 6) and confirmed the diagnosis of OPMD. His surgical repair was unremarkable, and within 1 week of surgery, the patient's back and neck pain began to improve. He was also able to resume his bicycling hobby. An otolaryngology consultation for his dysphagia was obtained.

One year following surgery, the patient's upper lid margin to reflex distance was 2 mm on each side with modest frontalis flexion (Figure 1D). He had a mild amount of lagophthalmos bilaterally. There was excellent contouring of his eyebrows with forehead flexion, and there was no evidence of neck extension. He reported complete resolution of his back and neck pain.

Case 2

A 12-month-old boy was referred to our Eye Plastics Clinic for bilateral severe blepharoptosis. He was born at full term by Caesarean section. His past medical history and family history were unremarkable. Per his parents, the patient had achieved his social, language, and cognitive developmental milestones, but had markedly delayed gross motor development, with poor crawling. On examination, he fixed and followed with each eye with no fixation preference and hence no evidence of amblyopia. The upper lid margin in primary gaze covered his pupils, with a margin to corneal reflex distance in each eye of -3 mm, and minimal eyelid excursion on each side. He had developed a significant chin-up head posture representing ocular torticollis (Figure 1E). His mother believed that this interfered with his ability to coordinate his movements and hence delayed his motor development. He underwent bilateral ptosis repair with silicone frontalis slings at the age of 13 months². At his 2-weeks post-op visit, his head posture was greatly improved, with minimal to no torticollis. At the 3-month evaluation, he fixed and followed equally with both eyes. Upper lid margin to reflex distance on each side was 4 mm with a trace amount of lagophthalmos; no torticollis was observed (Figure 1F). His mother was thrilled to report that within 1 month of surgery, he began to walk, and within 3 months had caught up with his gross motor development.

Case 3

A 4-month-old girl was referred for bilateral blepharoptosis. She was born at full term via normal vaginal delivery and the antenatal and other medical history was unremarkable. When seen in the Eye Plastics Clinic, she fixed and followed equally with both eyes. Her upper lid margin to corneal reflex distances were between 1 and 2.5 mm for both eyes, depending on frontalis flexion. She had mild telecanthus and phimosis, but no evidence of strabismus. She was diagnosed with blepharophimosis syndrome with moderate blepharoptosis. With no evidence of amblyopia, observation was recommended. She returned six months later, at the age of 10 months, at which time she presented with a marked chin-up head posture representing ocular torticollis. Her fixation was central, steady and maintained with no preference for either eye. The upper lid margin to reflex distances were 0-2mm depending on frontalis flexion, and eyelid excursion was less than 4mm. The horizontal palpebral fissure width was 14mm on right side and 13 on left side (Figure 1G). Her parents reported poor crawling and no cruising. Surgical repair was recommended,

which consisted of bilateral lateral canthoplasty and ptosis repair with silicone frontalis slings². Three months after uneventful surgery, she continued to have no evidence of amblyopia. The upper lid margin to reflex distances improved to 3 and 2.5 mm on right and left side, respectively. Furthermore, the ocular torticollis had resolved (Figure 1H) and she was now walking and meeting her developmental milestones.

Case 4

A 5-month-old boy was referred for evaluation of congenital ptosis and early preference for the right eye. His exam also revealed mild blepharophimosis and epicanthus inversus. His eyelid excursion appeared minimal, and his pupils were covered in primary gaze even with apparent maximal frontalis flexion. Whenever possible, the child extended his neck and lifted his chin in order to clear his visual axis (figure 1I), which his parents noted throughout the day. Based on the exam findings, bilateral upper lid ptosis repair with silicone frontalis slings was recommended. He underwent uncomplicated surgical correction, followed 4 months later by further tightening of the silicone sling on the non-preferred left side. Seven months later, his parents reported that his neck flexion was almost completely eliminated. His exam revealed good clearance of the visual axis (figure 1J), and his gross motor development was normal.

Discussion

Ocular torticollis is defined as an abnormal head position that maximizes visual acuity and maintains binocularity. Chronic neck extension with a chin-up position has been described in children with severe congenital blepharoptosis and in adults with severe acquired blepharoptosis¹. Williams *et al.*, in their study of 15 patients with torticollis, found neck tenderness in nearly half of the cases in the absence of true shortening or contracture of the sternocleidomastoid muscle³. They recommended that all patients with torticollis and no clear orthopaedic cause should be referred for an ophthalmic consultation. Similarly, Nucci *et al.*, in a multi-disciplinary study evaluating 63 children, found that about half of the children had abnormal head posture secondary to ocular torticollis⁴.

Patients with OPMD or other forms of myogenic ptosis, such as chronic progressive external ophthalmoplegia, or neurogenic ptosis, such as oculomotor nerve palsy or myasthenia gravis, may be more likely to assume a chin-up position compared to those with involutional blepharoptosis. This is most likely due to the severity of the blepharoptosis in these systemic diseases. In addition, CPEO and OPMD can also be associated with extraocular motility deficits including impaired supraduction, and a chin-up position may help maintain ocular alignment and binocular vision.

Our adult patient initially presented with debilitating back pain that was caused by ocular torticollis from blepharoptosis. The patient had undergone evaluations by multiple physicians, but the connection between his back pain and eyelid ptosis was not made. We present this case as an example of how chronic neck extension due to severe blepharoptosis can lead to back pain in an adult. Surgery in a case of myogenic blepharoptosis is delayed until the condition interferes with vision and/or day-to-day activities. This is mainly due to the risk of post surgical lagophthalmos and the risk of corneal complications from poor Bell's phenomenon⁵. The symmetric Blepharoptosis secondary to OPMD requires a conservative repair aiming to improve eyelid opening while minimizing lagophthalmos and blink abnormalities⁶. Our adult patient responded very quickly to proper surgical repair, with complete resolution of his back and neck pain, and maintenance of excellent vision. We have many additional patients, with less severe symptoms and findings, who still report months after surgery that they notice a marked improvement in neck and back pain that they had never attributed to their eyes.

Surgery in a case of severe congenital blepharoptosis is also traditionally delayed until preschool age unless there is an evidence of developing amblyopia. However, at least two of three pediatric cases in our series benefitted from early surgical intervention despite the absence of amblyopia, while the third had early evidence of mild eye preference, and severe neck flexion that was likely to interfere with development. To our knowledge there are no published studies that assessed gross motor developmental delays in children with severe blepharoptosis and ocular torticollis. Shapiro, in his review article, alluded to a relationship between head tilt and resulting tension on the lower back muscles in order to maintain posture⁷. Severe neck extension and chin-up position may thus interfere with gross motor development, which we observed in our patients. Whereas amblyopia can be assessed objectively, a delay in motor development is difficult to ascertain in the ophthalmologist's office, and would require direct questioning of parents. Importantly, in our patients, the parents reported improvement in their gross motor development immediately after surgical repair of the blepharoptosis.

Weaknesses of our study include its retrospective nature, and lack of formal neuromuscular evaluation. The latter is expensive, time consuming, and may be difficult to obtain, but may be helpful in certain cases as well as with future research studies.

In conclusion, we suggest that severe blepharoptosis is an unusual but important etiology of back pain in adults, and should be considered by physicians when evaluating patients. Similarly, children with severe blepharoptosis may be more likely to fall behind in their motor development. Ophthalmologists and primary care physicians should be aware of the systemic symptoms associated with severe blepharoptosis, since these can be addressed by correcting the eyelid position. Based on our observation of improved symptoms shortly after ptosis repair, we suggest that early surgery should be considered in cases of moderate to severe blepharoptosis that cause chin-up head posture and ocular torticollis. We also believe that further studies are needed to evaluate motor development in children with ocular torticollis.

Acknowledgments

This work was supported by a Research to Prevent Blindness Career Development Award to AK, and a Midwest Eye Bank Award to BLB. The sponsors had no role in the design or interpretation of the research. All the authors had full access to the data.

References

1. Rubin SE, Wagner RS. Ocular Torticollis. *Surv Ophthalmol*. 1986; 30(6):366–76. [PubMed: 3523807]
2. Carter SR, Meecham WJ, Seiff SR. Silicone frontalis sling for the correction of blepharoptosis: indications and efficacy. *Ophthalmol*. 1996; 103(4):623–30.
3. Williams CR, O'Flynn E, Clarke NM, Morris RJ. Torticollis Secondary To Ocular Pathology. *J Bone Joint Surg Br*. 1996; 78(4):620–4. [PubMed: 8682831]
4. Nucci P, Kushner BJ, Serafino M, Orzalesi N. A Multi-Disciplinary Study of the Ocular, Orthopedic, and Neurologic Causes of Abnormal Head Postures in Children. *Am J Ophthalmol*. 2005; 140(1):65–8. [PubMed: 16038652]
5. Wong VA, Beckingsale PS, Oley CA, Sullivan TJ. Management of myogenic ptosis. *Ophthalmol*. 2002; 109(5):1023–31.
6. Allen RC, Jaramillo J, Black R, et al. Clinical characterization and blepharoptosis surgery outcomes in Hispanic New Mexicans with oculopharyngeal muscular dystrophy. *Ophthal Plast Reconstr Surg*. 2009; 25(2):103–8.
7. Shapiro JJ. Relation between Vertical Facial Asymmetry and Postural Changes of the Spine and Ancillary Muscles. *Optom Vis Sci*. 1994; 71(8):529–38. [PubMed: 7970570]

Figure 1.

Ocular torticollis caused by blepharoptosis can be corrected with treatment of the ptosis. Children were photographed with their natural head position both before and after surgery. **A:** Example of neck extension caused by effort to maintain binocularity. **B–D:** Patient 1. **B:** Severe ptosis in primary gaze. **C:** Poor lid elevation in upgaze. **D:** Post-operative result with clearance of the visual axis through conservative elevation. **E–F:** Patient 2. **E:** Pre-operative photo showing severe ptosis and chin-up position to achieve axis clearance. **F:** Post-operative photo revealing improved lid and head position. **G–H:** Patient 3. **G:** Pre-operative photo showing severe ptosis and chin-up position to achieve axis clearance. **H:** Post-operative photo revealing improved lid and head position. **I–J:** Patient 4. **I:** Pre-operative photo showing severe ptosis and chin-up position to achieve axis clearance. **J:** Post-operative photo revealing improved lid and head position.