Introduction: Allen Putterman, MD (Attending)

This week’s grand rounds presents several interesting cases encountered by the oculoplastics service.

Schwartz-Jampel Syndrome: Erica Oltra, MD (Resident)

An 8 year old girl presented to the oculoplastics clinic with muscle stiffening of her arms and face that started around 2 years of age. She has had no progression of these symptoms throughout her life. Around 4 years of age, she was diagnosed with Schwartz-Jampel syndrome by genetic testing. Two of her sisters also have been diagnosed with this disease. Her past medical history was otherwise negative. Her social history was notable for the fact that her parents are second cousins.

On exam, she was noted to have “pursed” lips and bilateral blepharospasm, worse on the left (Figure 1). She had proximal muscle hypertrophy and thoracic kyphosis. Her vision was 20/60 OD and 20/60 OS. Her extraocular movements and pupils were normal in both eyes. She had a normal anterior and posterior ophthalmic exam.

DISCUSSION

Schwartz-Jampel syndrome, also known as chondrodystrophic myotonia, is a congenital disorder of the bone and muscle that causes a mask-like face, spinal misalignment, muscle hypertrophy, spontaneous muscle activity and extreme muscle stiffness. It also causes blepharospasm and ptosis. The disease has an autosomal recessive inheritance and has been found to be caused by a mutation of the HSPG2 gene, which encodes for the proteoglycan called perlecan. There are potential risks for general anesthesia in these patients including malignant hyperthermia and difficulties with intubation due to micrognathia.

Several treatments have been described for Schwartz-Jampel syndrome. Medical treatments include muscle relaxants and antiepileptic drugs such as carbamazepine. Local injections of botulinum toxin (Botox) are another treatment modality that may be used to treat the blepharospasm and facial spasm associated with this disorder. A few case reports exist in the literature describing surgical treatment with an orbicularis myectomy, levator aponeurosis resection and lateral canthopexy.

We treated our patient with periocular injections of Botox (total of 37.5 Units) and she has exhibited some improvement of her blepharospasm (Figure 2).
A Case of Monocular Proptosis: Elizabeth Grace, MD (Resident)

A 54 year old African American man was referred from his internist due to decreased vision and proptosis, or bulding, of the right eye. The patient noted the blurred vision and distance and near, and had been worsening over the last month. He also reports since late February 2011 his right eye started becoming more prominent, along with horizontal binocular diplopia on left gaze and a black spot in his vision on left gaze. He denies eye pain, no eye redness, no change in proptosis when he had a viral illness.

His past ocular history was notable only for myopic astigmatism for which he worse corrective spectacles. He denied eye surgeries, or eye trauma. His medical history was unremarkable, as he was healthy and on no medications. Family history notable for a brother with glaucoma. He works as a teacher, and doesn’t smoke, drink or use illicit drugs. Review of systems was completely negative.

On exam, his right eye was only correctable to 20/40 with -5.25 +2.75 x090; and the left eye was correctable to 20/20 with -4.00 +2.75 x085. Ishihara color vision was 11/11 in right and left eyes. The right eye was remarkable for 2+ resistance to retropulsion and proptosis OD, with Hertel measuring 34 OD and 28 OS, base 114. His motility OS was full, and OD was restricted -1 to -2 in all fields of gaze. He had no RAPD in either eye, confrontational visual fields were full OU, and IOPs were 20 OD and 19 OS. Eyelid measurements showed, palpebral fissure 13 OD versus 9 OS, MRD1 5 OD versus 3 OS, scleral show 4 OD and -2 OS, and symmetric levator function 15mm OU. The anterior SLE was unremarkable OU, and the dilated fundus exam showed a cup/disc ratio 0.3 OU with no disc edema, no disc pallor, a sharp foveal reflex, and otherwise an unremarkable peripheral exam OU.

A work-up was obtained, which was negative for thyroid dysfunction, and CT scan of the orbit showed a well encapsulated oval shaped mass in the retrobulbar space of right orbit, abutting globe and displacing the optic nerve (see Figure 3). To further differentiate the lesion an MRI of the orbits was performed, and showed a multiloculated cystic lesion filling most of the intracanal space (see Figure 4 and 5). The lesion had an enhancing capsule and some hypointense layering fluid levels thought to be blood, and thinning of the orbital roof, though without frank extension into the cranium. At this point the differential diagnosis was cavernous hemangioma, versus lymphatic or AV malformation, versus cyst, versus metastasis or other.

The lesion was fortunately not causing an optic neuropathy, however the nerve was on stretch. Because of this increasing proptosis, diplopia on side gaze, and his cosmetic appearance the patient decided to have surgery to debulk the tumor and to obtain a tissue diagnosis of the lesion. The patient underwent a right lateral orbitotomy with removal of the lateral orbital wall. After removal of the lateral orbital wall, a thinned orbital roof was identified, and the lesion was identified as pink cystic structure filled with serosanguinous fluid (see Figure 6). The lesions was debulked, while pieces of the capsule were sent for frozen path, and returned as non-specific fibrous tissue. The final path came back the next week as a schwannoma, with positive immunohistochemical staining for S-100.

DISCUSSION There are a variety of surgical approaches to an orbital tumor, however most importantly is the location of the tumor as this usually dictates the approach. In this gentleman’s case three options were discussed with him due to the highly posterior nature of the tumor, and the large intracanal size of the mass. The best exposure of the tumor would have been through a superior approach, or craniotomy with neurosurgery. This would also provide the greatest opportunity to remove the lesion in its entirety, although without guarantee and with the greatly increased side effects that can occur from a craniotomy.
A Case of Monocular Proptosis:  (continued)

DISCUSSION (continued) Another idea was through interventional radiology, which would be the least invasive, however would only provide a biopsy for diagnosis and not likely provide any “debulking” of the large lesion. Interventional radiology, however did not offer the patient any procedure and thus he chose the lateral orbitotomy.

Interestingly schwannomas are benign slow growing tumors that originate from the Schwann cells found along the peripheral and sympathetic nerves. Orbital schwannomas typically occur in young to middle aged adults and present as a well-circumscribed mass, located in the intraconal or extracranial space. On MRI they are usually isointense to extraocular muscles, however as in our case they may undergo necrosis and present with a cavitary or cystic appearance. Pathologically these encapsulated tumors can form solid structures (Antoni A pattern) or a looser more myxoid appearance (Antoni B pattern). Ideally treatment is by total excision, to prevent recurrence and the extremely rare malignant transformation. This patient’s tumor could not be completely debulked due to tumor infiltrating some large orbital veins, thus he will have future surveillance MRIs to monitor for recurrences.

References:

Optic Nerve Sheath Biopsy:  Mark Krakauer, MD (Resident)

A 23 year old man who was referred to the neuro-ophthalmology clinic at UIC by an outside neurosurgeon for vision loss. He had a history of transient visual obscurations 8 months prior and was found to have bilateral ON edema, normal visual acuities, and visual fields with enlarged blind spots on exam 6 months ago. MRI at that time showed a small left thalamic lesion and enlarged optic nerve sheaths. He was treated with dexamethasone for 3 weeks. Serial MRI’s were stable, but recently the patient noticed his vision would gray out upon standing up. 12 days prior he developed fatigue and malaise, his vision worsened, and he had a bilateral field cut in the lower left periphery. He was started on dexamethasone 4 mg PO BID. On exam, his best corrected visual acuity was 20/50 OD, 20/200 OS, his color vision was 11/11 OD, 6/11 OS, and he had severe b/l optic disc edema extending to macula OU. He was started on IV solutedrol 1g x 3 days and Diamox 500mg PO BID, and an LP and lab workup for inflammatory causes of optic nerve edema was ordered.

The MRI showed a left thalamic lesion with minimal mass effect on the 3rd ventricle, and optic nerve sheath enhancement (Figures 7 and 8). The LP was normal, as were lab tests, and additional radiological studies looking for malignancy or sarcoidosis. The left thalamic lesion and dura were biopsied by neurosurgery, and pathological analysis showed a grade 3 anaplastic astrocytoma. The leptomeningeal biopsy showed atypical glial cells with pS3 staining.
Optic Nerve Sheath Biopsy: (continued)

Dural biopsy was negative for tumor. During the course of his hospital stay his visual acuity went from 20/50 to CF at 2 feet OD, and from 20/200 to LP OS. Additionally his color vision and visual fields deteriorated despite IV steroids. It was unclear if his vision loss was due to carcinomatous meningeal involvement of the optic nerve sheaths. If the malignancy had not spread down the optic nerves then the patient could receive focal radiation therapy. However, if the malignancy involved the optic nerve sheaths, then he would require whole brain radiation therapy (Koukourakis). An optic nerve sheath biopsy was performed on the left eye via a subconjunctival approach, requiring disinsertion of the medial rectus in order to gain access to the deep intracanal space and the optic nerve. Pathological analysis of the optic nerve sheath showed p53 staining in a few cells. This was supportive, but not diagnostic of leptomeningeal involvement in the optic nerve. The patient was started on whole brain radiation and chemotherapy and after 1 week the vision improved to 20/400 OD, HM 4’ OS.

DISCUSSION There are few studies looking at optic nerve sheath biopsy. In one case series, at the University of Pennsylvania, over 20 years, 18 patients had optic nerve biopsy of intrinsic or adherent masses for tissue diagnosis (Levin). Only 2 eyes gained or lost substantial vision, and in only one case was a clinical diagnosis not reached. Another case series of 86 patients who had optic nerve sheath fenestration for idiopathic intracranial hypertension showed only one long-term complication from the surgery (Banta). Overall, taking a biopsy of the optic nerve sheath is an extremely rare procedure, although it is performed technically similar to a optic nerve sheath fenestration in terms of exposing the optic nerve. The procedure can be technically difficult due to the difficulty of accessing the cone, as well as taking a large enough sample for pathology without damaging the optic nerve or its blood supply.

References:

UPCOMING CME COURSES

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Upcoming Illinois Eye and Ear Infirmary Ophthalmology Grand Rounds are held Wednesdays at 5:00 pm on the UIC campus at 909 S. Wolcott in the College of Medicine Research Building. For a complete schedule go to www.uic.edu/com/eye and click on Grand Rounds under the Education drop down menu. Or, call 312-996-6590.