Recent Onset Ptosis and Proptosis in an Infant with Neurofibromatosis Type 1

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Abstract:

This case report discusses the ocular manifestations of neurofibromatosis in the setting of a patient that presents to clinic with a recent onset ptosis and undergoes imaging to reveal a plexiform neurofibroma of the orbit.

Case History:

An 11 month old Caucasian male presents to clinic because his mother notes a ptosis of the left eye that began three months ago. The patient’s mother has no other concerns; the patient displays age appropriate visual behaviors. His systemic history includes a diagnosis of neurofibromatosis type 1. In addition, the patient has a strong family history of neurofibromatosis type 1, which includes both his mother and older brother.

Pertinent Findings:

The patient is able to fix and follow a toy in both eyes. His pupils are equal, round and reactive to light in both eyes, and there is no afferent pupillary defect. Confrontation fields are grossly full to distraction. External observation of the patient reveals a slight proptosis of the left eye, without a palpable periorbital mass. There is a ptosis of the left eye that does not obscure the visual axis. The MRD1 in the right eye is 5 mm as opposed to an MRD1 of 2.5mm of the left eye. Slit lamp examination is normal and without Lisch nodules in both eyes. Intraocular pressure by icare tonometry is 9 mmHg in the right eye and 11 mmHg in the left eye. Cycloplegic retinoscopy reveals anisometropia of +2.50 +0.25 X 090 in the right eye and -1.50 +1.50 X080 in the left eye. Dilated ocular health is unremarkable in both eyes.

Differential Diagnosis:

Differential diagnosis includes optic nerve glioma and plexiform neurofibroma.

Radiology Studies:

Magnetic resonance imaging shows increased abnormal signal in the left orbit, with involvement of the intraconal and extraconal soft tissues, and extension into the lacrimal gland. There is remodeling of the left sphenoid triangle/lateral wall of the left orbit. The imaging reflects a plexiform neurofibroma of the left orbit. Enhancing soft tissue is also noted at level of foramen ovale as well as the pterygopalatine fissure, which may reflect neurofibromas or additional plexiform neurofibromas.
Diagnosis and Discussion:

Neurofibromatosis type 1 is an autosomal dominant condition caused by a mutation in the neurofibromin gene on chromosome 17. It occurs in 1 of every 3500 people, and approximately half of cases are due to spontaneous mutation. Signs of neurofibromatosis type 1 include café au lait spots, neurofibromas, freckling in the armpits or groin, and bone deformities such as scoliosis. Individuals with neurofibromatosis type 1 may also have a shorter stature, large head size, learning disabilities and cardiac problems. The ocular manifestations of NF1 include lisch nodules on the iris, optic nerve glioma, and plexiform neurofibroma.

A plexiform neurofibroma is a hamartoma of neuroectodermal origin that comprises approximately 1-2% of orbital tumors. Plexiform neurofibromas are less common than optic nerve glioma in patients with neurofibromatosis. These tumors typically arise during the first decade of life and involve the sensory nerves of the orbit and eyelid. Plexiform neurofibromas may cause an “S” shape to the eyelid due to thickening and fat deposition. In more advanced cases the sphenoid bone becomes involved which can lead to proptosis and bony expansion. Secondary ocular complications include deprivation, refractive, and strabismic amblyopia. One study found that 60% of children with a plexiform neurofibroma have amblyopia in the affected eye. In rare cases the tumor may become malignant, and half of patients with a plexiform neurofibroma will develop an ipsilateral glaucoma secondary to angle closure.

Treatment and Management:

Enlargement of plexiform neurofibromas during childhood and adolescence can result in facial disfigurement. Treatment is aimed at alleviating gross disfigurement and deprivation amblyopia. Complete surgical resection is often not possible because the tumor is unencapsulated, highly vascular and infiltrative. Visual prognosis is dependent on the size and location of the tumor. Patients should receive a complete eye exam every six months which must include visual acuity, motility, intraocular pressure measurements, a cycloplegic refraction, color vision and stereopsis. Frequency of visits should be increased if there is a decline in vision or growth of the tumor.

The patient discussed in this case will return one month after initial presentation for a follow-up cycloplegic refraction. At this time glasses will be prescribed for full time wear to treat potential refractive amblyopia. The patient will be carefully observed for tumor growth by clinical exam and serial imaging.

Conclusion:

A multidisciplinary team is necessary to manage patients with ocular manifestations of systemic conditions, such as neurofibromatosis. It is crucial to address the basic optometric needs of patients which include treatment of amblyopia and the ocular surface, along with coordinating care between other providers. Although the visual prognosis for patients affected
by NF1 is unpredictable, it is essential to maintain a patient-centered approach when caring for all patients.

References: