

## **Pediatric Ocular Disease (revised)**

### **Abstract:**

This case report is of a 1 year old who presented with a neurofibroma. His progress is tracked over a period of 5 years. The initial ocular lesion was a tumor in the superior lateral aspect of his left upper lid. Patient management included the initial monitoring of the condition and eventual surgical intervention. The ocular and visual manifestations included a continued enlargement of the tumor with alterations to the anterior segment and the development of myopia and reduced visual acuity.

### **Introduction:**

Neurofibromatosis presents clinical as two distinct autosomal dominate diseases. Type 1 (NF-1) has an incidence of 1 in 3,000 has been referred to as Von Recklinghausen Disease. Type 2 (NF-2) is far more rare with an incidence of 1 in 50,000. The clinical presentation helps to differentiate type NF-1 from NF-2. A diagnosis of NF-1 can be made if two of the following can be made: six or more café-au-lait spots of over 5 mm in diameter, two or more neurofibomas, one plexiform neurofibroma, freckling in the axial or inguinal regions, optic nerve glioma, two or more Lisch nodules, or a first degree relative having the condition. A diagnosis of NF-2 can be made if there is a bilateral eighth nerve mass as seen by CT or MRI testing, or a first degree relative who has a unilateral eighth nerve mass or two of the following: neurofibroma, meningioma, schwannoma, or a juvenile posterior subcapsular lens opacity.

## **Case Report:**

### **Initial Examination:**

Nathan was first seen (5/11/94) when he was 1 year 2 months old. The chief complaint was the presence of an occasional red left eye, which was accompanied by tearing, and matting of the lids. The child was an 8 lbs. 11 oz. full term baby, born to a mother who was over 35 years of age. Pre, peri and postnatal history were unremarkable. At birth Nathan was healthy and required only routine postnatal care. Nathan was one of a number of children, and though the mother could not remember exact developmental milestone dates, she felt that they occurred within expected limits. The one exception was that Nathan had yet to walk unaided. The child was healthy and was not taking any medicines. He had no known allergies. Family ocular history included a number of family members who were myopic. Family health history was unremarkable.

Following this history, a comprehensive examination was done. Visual acuity (VA) was taken with both eyes open at 55cm using Teller Cards. The VA was 20/60. Cover tests at both distance and near were ortho. Near point of convergence (NPC) was 2 inches. There were no extra ocular muscle limitations, and the eye movements were full and concomitant. Nathan passed plates 6 and 9 on the Keystone Basic Binocular series. Dry and cycloplegic refraction was +1.50 diopter sphere right eye (OD) and left eye (OS). The color, brightness and stability of the reflexes were equal. Pupils were equal, round and reactive to light and accommodation. There was no afferent pupillary defect. External examination revealed a clear OD; however the OS presented with a matted lid with some conjunctival edema and chemosis of the lower lid. The nasolacrimal duct of the OS was obstructed. This was determined by the regurgitation of matter when the

nasolacrimal duct was palpitated. Also noted was a general puffiness of the superior temporal lid of the left eye. The area was soft and not tender. Digital tensions were soft and equal. Dilated fundus evaluation was unremarkable. In each eye the media was clear. Cup/discs (C/D) were 0.1 and round. The discs were pink, round and well defined. Artery/vein (A/V) ratios were 2/3. The central retina of each eye was clear. The peripheral retina of each eye was flat and without holes. At the end of the examination the mother was further questioned about Nathan's general health. At this point she reported that the pediatrician had noted a number of café au lait spots; however, she hadn't thought it was a pertinent part of his health history. In addition, she had no knowledge of anyone in the family having these spots.

The overall assessment was that of a child who had normal visual development. VA and refraction was age appropriate. His eyes were aligned with gross stereopsis. Though not walking yet, the child's motor development when using Shirley's norms for posture and locomotion (University of Minnesota) of 15 months, appeared within normal limits. The fundus grounds were negative. The conjunctival congestion was secondary to a congenital nasolacrimal duct obstruction. However of greater concern was the superior temporal mass in the left lid. This coupled with the added history of café au lait spots increased the concern of a possible neurofibroma.

The following is the treatment and management plan. Nathan's mother was advised to keep the eye clear of debris. She should use warm compress and massage the OS nasolacrimal duct in a forceful downward manner 10 times a day for a period of a minute each time. However since the child was over a year old and the condition had not spontaneously resolved, there was increased likelihood that probing would be necessary

for a better outcome. Of more concern was the potential mass in the superior temporal area of the OS. The patient was referred to an ocular plastic surgeon for further evaluation of both conditions. The mother was also advised that a MRI and CT scan maybe ordered by him, as well as a possible, neurological consult to rule out a neurofibroma. A report would also be sent to the child's pediatrician, so that he might be a coordinator in this effort. Nathan was to return for a progress evaluation in 3 months.

**Subsequent Visits:**

The following are a summary of the salient points of subsequent visits:

Nathan was next examined on 8/18/94. At this time, Nathan hadn't had any imaging done. His mother was in the process of following through on the previous recommendations. The OS lid appeared to have increased in fullness and thickness. The ocular alignment and refraction remained unchanged from the previous visit. The mother was advised to follow through on the previous recommendations. Nathan was to return for a progress visit in 3 months. This time frame was used to serve two purposes, i.e. to monitor Nathan's condition and to make sure of the continuation of care at the secondary level.

Nathan next visit was on 12/12/94. A MRI and CT scan had been performed. The diagnosis of neurofibromatosis had been confirmed. At this time no surgery was recommended. Other ocular findings remained unchanged. He was to return in 3 months.

Nathan next examination was on 2/28/95. There was a continued increase in the fullness and thickness of the lid. The characteristic S shape lid was evident and the mass was starting to increase into the nasal aspect of the superior lid. The refraction in the OS had shifted into low myopia, -1.50 diopter sphere area. The cornea of the left eye was

clear. However the impression was that this increase in the mass was causing an increase in overall length of the eye, and thus the emergence of the myopia. No spectacle prescription was given at this time. Nathan previous visual development had been normal, thus there was no great concern about correcting his change in refractive status. He was to return in 3 months.

Nathan's next visit was on 6/12/95. The mass appeared to be more stable. Photos were taken at this time. (They didn't reproduce adequately enough – thus not included. The originals were very dark and not the greatest). The other findings were unchanged. He was to return in 3 months.

The next two visits were on 11/2/95 and 2/26/96. The mass seemed relatively stable. The myopia finding in the OS remains in the -1.50 diopter sphere area. He was to return in 3 months.

Nathan's was next examined on 5/13/96. The mass had increased in size and now involved the lower temporal lid. At this point the mass had involved the full superior lid and half of the inferior lid. Corneal warpage was also noted at this visit.

The patient had continued follow up care with both the ocular plastics ophthalmologist and the neurologist. On 6/17/97, surgery was done. The vast majority (90% area) of the mass was removed. In this case, it is understandable that the surgeon put off surgery, for recurrence and unsuccessful outcomes with poor prognoses are unfortunately the usual result. Also of major concern is the fact that these lesions may arise throughout the central nervous system.

Nathan was examined on 12/1/97. Nathan presented with a marked increase in myopia and a variable correctable VA finding in the OS, which ranged from 20/40 to

20/60. The refraction was in the  $-4.50$  area. The OD remained in the emmetropic area. Dilated fundus exam was negative. In order to verify or duplicate these refractive changes, the patient was scheduled for a two-week follow-up visit. There was also some question as to whether there was some thickening of the lower temporal aspect of the lid. On 12/17/97 Nathan returned. At this visit, the previous findings were duplicated. The correctable VA was 20/30. The reduction in correctable VA was secondary to some of the corneal warpage. In order to maintain some size consistency, Nathan was prescribed a  $-3.00$  diopter sphere for the OS (polycarbonate for safety). Though Nathan condition was probably due to the alteration of his cornea and not amblyopia, it was felt that patching therapy would do not harm. He was instructed to direct patch for 2-3 hours a day. Due to the mass size, it's alteration of lid aperture, corneal warpage, and the need for safety protection; contact lenses were not prescribed.

Nathan's next visit was on 2/9/98. His findings were similar to his last visit. There was some thickening of the upper and lower lid. The treatment plan was not changed.

Nathan's next visit was on 4/27/98. Nathan had not been very compliant with patching. There was an increasing fullness to both the upper and lower lid. The involvement had extended to the medial aspects, and the superior and inferior cornea was being compressed. Corneal distortion was evident on keratoscopic evaluation. In addition the blink mechanism had been compromise, such that, there was not full coverage of the corneal surface. The refraction had now stated to display a minus cylinder at 90. Artificial tears were prescribed tid to qid or prn. There was no change in the spectacle Rx. Though there are alternatives to patching non-compliance, i.e. penalization or fogging; the child's overall well being needed to be considered. Thus, no alternative was invoked.

The next two visits were on 5/14/98 and 8/17/98. There were similar findings as the last visit, except there was some increase in the general overall fullness of the mass.

Nathan's next visit was on 2/1/99. Nathan hadn't patched. His intraocular pressure (IOP) was 16 OD and OS. Fundus evaluation was negative. Confrontation fields were negative. EOM remained full. The mass had increased in all aspects and was encroaching onto the temporal cornea. The OD iris presented with a nodule at 8:00 o'clock. There was a slight increase in myopia but the VA was stable. He was to continue with his present glasses. The plan was to consult with the surgeon again.

Nathan was next seen on 5/10/99. There continued to be an increase in size of the mass and its encroachment onto the cornea. VA OS had dropped to 20/60. Due to the poor prognosis, the surgeon preferred not to repeat the surgery.

Nathan's next exam was on 2/28/00. There was further increase of the mass, such that there was a restriction of left lateral gaze. There was also the presence of a left hypotropia. The OD iris nodule remained as noted previously. At this time the neurologist and the surgeon preferred not to repeat a MRI, nor to do surgery. The child's parents are considering an alternative treatment project at the University of Pittsburgh.

### **Discussion:**

Nathan's condition is a NF-1 type of neurofibromatosis. Surgical resolution of the ocular manifestations is as a rule unsuccessful. However, the lesions may be disfiguring and at times surgical intervention is persuaded. Cerebral, brainstem, and cerebella tumors may also arise. Neurological imaging is initially used to delineate any central nervous system lesion. Subsequent imaging is usually reserved for unexplained neurological or

ophthalmic presentations. The ocular tumors can involve the optic nerve, and even progress posteriorly toward the chiasm. However medical treatment is frequently more monitoring than active intervention, which is reserved for more profound presentations of the disease. Surgical as well as radiological and ontological treatment modalities have been used.

Over the duration of Nathan's care, his ocular tumor has been excised only to reoccur at an even more dramatic rate and degree. The growth has resulted in facial disfigurement. Psychological intervention has helped this very nice young man deal with his condition. Presently, the growth has compromised his corneal topography, his eye movements, and most likely his optic nerve. Nathan's reduced VA is more attributable to pathology than the development of amblyopia. It would appear as though the growth has altered the shape of his left eye to render it myopic. Compression on the side of the globe could theoretically lengthen the anterior-posterior measurement. If the compression were from directly behind the globe, it would likely alter the eye in a hyperopic direction.

Nathan was routinely seen on a three month basis. This time frame could have been expanded, but by doing 3 month recalls, he stayed in the health care loop. In addition, the ocular tumor presentation was the most overt component of the disease, and in this case, the optometrist served as a coordinator of care.