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MACULAR DETACHMENT ASSOCIATED WITH ANOMALOUS OPTIC NERVES AND DURAL ECTASIA IN 49, XXXXY SYNDROME

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Abstract

Purpose—To present a case of a patient with XXXXY syndrome, anomalous optic nerves, and dural ectasia in conjunction with macular detachment.

Methods—Case report.

Results—A 3-year-old boy with XXXXY chromosomal abnormality presented with bilateral maculopathy. On evaluation, he was found to have anomalous optic disks with serous detachment of the left eye. Magnetic resonance imaging of the brain revealed bilateral optic nerve dural ectasia without evidence of elevated intracranial pressure.

Conclusion—XXXY syndrome, like the related condition of Klinefelter syndrome, can manifest with ocular abnormalities. In the present case, the dural ectasia may have facilitated access of cerebrospinal fluid through anomalous optic nerves, resulting in neurosensory detachment.

Keywords

disk anomaly; serous retinal detachment; XXXXY syndrome

Case Report

A 3-year-old boy with 49, XXXXY syndrome was referred for the evaluation of a macular dystrophy. He was referred by his pediatric ophthalmologist who had seen him for persistent right eye tearing, which resolved after tear duct probing. There were otherwise no ocular

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symptoms, and his parents did not have any concerns about his vision. His medical history was notable for delivery by cesarean section in the setting of preterm labor at 30 weeks of gestation, 49 chromosomes with the XXXXY karyotype, small size, hypertelorism, camptodactyly, clinodactyly, large ears, Arnold Chiari Type I, and developmental delay. No family members had ophthalmic disease or chromosomal abnormalities. On examination, vision was 20/100 in the right eye and the left eye. Cycloplegic refraction was -0.50 sphere in the right eye and plano sphere in the left eye. Ophthalmoscopic examination revealed anomalous nerves with bilateral retinal pigment epithelial changes tracking from the optic nerve to the macula in each eye. Optical coherence tomography revealed subretinal fluid in the left macula (Figure 1). Optical coherence tomography of the optic nerves revealed enlarged optic nerve cups, vitreopapillary traction in both eyes, and a peripapillary intrachoroidal cavitation at the inferonasal edge of the optic nerve in the left eye (Figure 2). Review of the magnetic resonance image revealed enlargement of the supratentorial ventricular system, enlargement of the lateral ventricles, a Chiari Type I malformation, and an increased fluid signal around both optic nerves, compatible with optic nerve dural ectasia. Cerebrospinal fluid pressure was normal.

Discussion

The 49, XXXXY syndrome is caused by the presence of three additional X chromosomes in a male. Occurring in approximately 1:100,000 males, it has some shared features with Klinefelter syndrome (47, XXY) but more severe mental, physical, and psychological manifestations. Previous studies have reported associated ocular anomalies including hypertelorism, epicanthic folds, and high myopia.¹ A patient with Klinefelter syndrome and a unilateral optic pit with associated macular serous detachment has previously been reported.²

In 1882, Wiethe described the first case of bilateral optic pits. Histologic assessment of optic pits consisted of a congenital defect in the closure of lamina cribrosa, resembling a small coloboma of the optic nerve.³ Optic disk pits can be asymptomatic,⁴ but they can also commonly present with macula-related complications like serous retinal detachment.⁵ The pathophysiology of serous macular detachment in optic disk pit remains controversial.^{6,7} Previous studies have suggested that one potential source may be cerebrospinal fluid from the subarachnoid space.

Interestingly, this patient had bilateral dural ectasia of the optic nerve found on magnetic resonance imaging. Optic nerve dural ectasia is the enlargement of the optic nerve sheath as a result of the increased flow of the cerebrospinal fluid without a corresponding increase in intracranial pressure. In this case, the dural ectasia may have facilitated access of the cerebrospinal fluid through anomalous optic nerves, resulting in the neurosensory detachment.

Other etiologies could have also contributed or even caused these findings but may be less likely in the context of this patient. Zumbro et al⁸ reported macular detachment in subjects with enlarged optic nerve heads because of acquired glaucomatous cupping. None of the subjects in their series had congenitally anomalous nerves. Vitreopapillary traction has been

described in other subjects with concomitant macular detachment as a result of vitreomacular traction,⁹ which was not present in this case (Figure 1). Peripapillary intrachoroidal cavitation has been associated with macular detachment in high myopes,¹⁰ but in these subjects, the cavitations were most commonly large, temporally located, and had evidence of contiguity with the retinal detachment all of which are lacking in this case. Moreover, the macular findings in conjunction with the anomalous nerves and dural ectasia of this case are bilateral, but peripapillary intrachoroidal cavitation was only evident in the left eye. Finally, eyes with pathological myopia can have an associated macular schisis and detachment, but this patient was not highly myopic.

In summary, we present a case of XXXXY syndrome in which macular detachment is associated with dural ectasia and anomalous optic nerves.

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Fig. 1.

Color photograph of disk and macula of both eyes (**A** and **B**). **A.** Right eye reveals anomalous optic nerve with enlarged cup. Retinal pigment epithelial hypopigmentation and atrophy as well as pigment migration into the retina are evident tracking from the nerve to the fovea. **B.** Left eye also reveals retinal pigment epithelial hypopigmentation, atrophic change, and pigmentary clumping that extends from the nerve to the macula. Inferonasally, a peripapillary intrachoroidal cavitation is evident. **C.** Optical coherence tomography of macula demonstrates retinal pigment epithelial atrophy in the nasal macula with disruption and some loss of the inner segment ellipsoid band. **D.** In the left eye, optical coherence tomography reveals a neurosensory detachment in the macula again with nasal retinal pigment epithelial atrophic changes and attenuation of the inner segment ellipsoid band.

Fig. 2. Optic nerve in the right eye shows an enlarged optic nerve cup (dashed circle) (**A**) and optical coherence tomography (arrow) (**B**). The left optic nerve is anomalous with both temporal and nasal cupping evident (dashed circles) (**C**) and arrows on Optical coherence tomography (**D**). Centrally dislocated vessels are present, producing posterior shadowing with associated vitreopapillary traction. Nasally, a small peripapillary intrachoroidal cavitation is evident (**D**).