Beyond refractive error: A sibling case of chorioretinal coloboma mimicking astigmatism

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ABSTRACT

Introduction: Chorioretinal coloboma is one of the rare congenital ocular malformations that result from defective closure of the embryonic fissure during the early weeks of gestation. The condition may cause visual problems, depending on the location of the tumour and its size, with possible complications such as retinal detachment or choroidal neovascularisation. The disease is usually diagnosed at an early age even if visual impairment is severe, but cases are sometimes only detected when it is too late and the child has already attended school, for instance, presenting as refractive errors. In addition, familial cases have rarely been reported, and siblings presenting with high astigmatism have not been described. **Case description:** Two siblings presented to the ophthalmology clinic with complaints of blurred vision and high cylindrical refractive errors. The first case, a 9-year-old girl, had anisometropia with a cylindrical refractive error of –4.00 dioptres (D) in the left eye and –0.50 D in the right. Her 14-year-old brother had –5.00 D in the right eye and –0.50 D in the left. Both had normal anterior segment findings. Fundoscopic examination revealed inferonasal chorioretinal colobomas in both siblings. The brother's left eye also demonstrated optic disc and macular involvement, with sharply demarcated areas of absent retina and choroid, exposing underlying sclera. Optical coherence tomography confirmed deep excavation corresponding to the coloboma margins. No systemic abnormalities were present in either patient, and there was no family history of ocular disease. Both patients were prescribed corrective lenses and scheduled for routine follow-up to assess visual function and identify any complications. **Discussion:** These cases illustrate that chorioretinal coloboma can present with high astigmatism due to irregular retinal structure, rather than corneal deformity alone. The bilateral findings in siblings raise the possibility of an underlying genetic predisposition, we suggest that genetic counselling and screening of at-risk relatives should be done. Children with high astigmatism and anisometropia are at a substantially increased risk of developing amblyopia; therefore, early screening and timely refractive correction are essential to optimise visual outcomes. Moreover, the lifelong risk of retinal detachment in patients with coloboma emphasizes the importance of patient and parent education regarding alarm symptoms, including sudden vision loss, flashes, or floaters. The findings in this and similar cases support fundus examination as part of the ocular workup whenever a child presents with significant unequal or bilateral refractive errors. **Conclusion:** Chorioretinal coloboma may sometimes present in children as high or asymmetric astigmatism, without other obvious ocular and systemic signs. It is important to consider this condition, particularly in cases of unusual refractions within the paediatric population. Early detection promotes timely referrals for refractive correction and reduces the likelihood of amblyopia, while also aiding in the identification of potential sight-threatening issues, such as retinal detachment. Even in the absence of familial occurrences, genetic counselling and screening of relatives can be beneficial for planning clinical interventions and ensuring long-term monitoring.

Keywords: Chorioretinal Coloboma, astigmatism, anisometropia, Pediatric Ophthalmology