Ophthalmology and Eye Diseases

SHORT REPORT

Peters' Anomaly

Robert W. Sault[1] and Jeffrey Sheridan[2]

[1]Biomedical Research, Kelowna, BC, Canada. [2]Board Certified Ophthalmologist Total Eye Care Center, Clermont, Florida, USA. Corresponding author email: rwsault@shaw.ca

Abstract: While conducting medical aid in Mozambique, a 41 year old African male presented to our eye clinic complaining of visual impairment. The male was found to have Peters' anomaly type 2, a rare congenital ocular malformation leading to sensory amblyopia and glaucoma.

Keywords: Peters' anomaly, genetic disorders, eye diseases, sensory amblyopia, ocular malformations

Ophthalmology and Eye Diseases 2013:5 1–3

doi: 10.4137/OED.S11142

This article is available from http://www.la-press.com.

Short Report

While conducting medical aid in Mozambique, a 41-year-old African male presented to our eye clinic with long-standing visual impairment beginning in early childhood. He had no additional ophthalmic or systemic symptoms and reported being otherwise well, including a review of systems that was negative for any findings. Past medical history included multiple respiratory infections in childhood, one case of acute otitis media without treatment, and no surgical history. The patient denied current use of any medications or over-the-counter products. Family history reported positive for cataracts on the paternal side; the maternal side was unknown. Social history elicited light tobacco and no alcohol consumption. Physical examination revealed a male of short stature with dense bilateral cataracts (*white arrows*) and bilateral adhesions of the iris to the posterior cornea (*black arrows*). Loss of visual acuity was significant, with the patient detecting only light and hand motion in front of his face. Intraocular pressure taken undilated with a Tonopen measured 24 mmHg bilaterally. Gonioscopy noted open angles with difficult visualization of the trabecular meshwork.

A differential diagnosis for this patient would include:[1] congenital cataracts, corneal graft rejection, posterior polymorphous corneal dystrophy, and sclerocornea. However; in lieu of the physical examination revealing dense bilateral cataracts, bilateral iridocorneal adhesions in conjunction with a short stature, these findings are consistent with Peters' anomaly, a very rare autosomal recessive ocular malformation that can be associated with systemic abnormalities. Systemic abnormalities include: cleft lip and palate, facial dysmorphism, syndactyly, brachycephaly, as well as cardiac, neural, and hearing deficits.[2] Most cases are sporadic or recessive; however, there are reports of dominant inheritance in the literature.[3] The incidence and prevalence of Peters' anomaly internationally remains unknown.

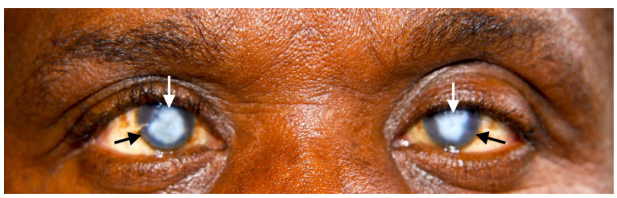

Figure 1. Photograph by Robert W Sault—undilated.

Most common gene mutations include PAX6[4] and FOXC1,[5] noting these genes are responsible for ocular embryogenesis. Peters' anomaly can be classified into two subtypes: Type 1, and Type 2. In Type 1, 80% of cases present bilaterally with central and paracentral corneal opacification. The cornea is typically avascular; iris strands extend from the collarette, and systemic abnormalities are not usually present. In Type 2, cases are commonly bilateral with denser corneal opacification; there is often juxtaposition of the lens, and iris strands may or may not be present.[12] The posterior stroma and Descemet membrane is classically malformed.[6] The incidence of systemic abnormalities is more common in Type 2.[7] In keeping with the examination findings of the patient, we classify this case as Type 2 Peters' anomaly.

Sensory deprivation, amblyopia, and glaucoma are significant sequelae of Peters' anomaly.[8] It is vital for the ophthalmologist to classify it as either Type 1 or Type 2, and to receive multiple consultations from neonatology and genetics pending a diagnosis following birth.[9,10] A later diagnosis recommends that patients undergo molecular genetic testing and receive consults from retina, cornea, and glaucoma specialists. Guidelines recommend the initiation of treatment as early as possible in order to prevent sensory amblyopia.[11] While Dorzolamide is available to treat pediatric glaucoma, early surgical intervention is strongly advised.[12–15] In a patient presenting with a clear peripheral cornea, peripheral iridectomy is the treatment of choice. Among patients presenting with bilateral corneal opacity and significant visual impairment, penetrating keratoplasty remains the standard of care; additionally, pooled analyses indicate rates of graft clarity between 20%–60%.[16–19] In patients with cataract alone, the treatment of choice remains lensectomy and vitrectomy.[20] Due to limited resources, our patient received extensive counseling in the medical management of glaucoma and was discharged from our clinic in good spirits.

Acknowledgments

The authors wish to acknowledge the generosity of the patient and his family for consenting to participate in this case report. Additionally, the authors wish to thank the people and hospital staff of Macomia, Mozambique for their hospitality.

Funding
Author(s) disclose no funding sources.

Author Contributions
Robert W. Sault is first and corresponding author of this paper. The patient's diagnosis within this case report was made by Jeffrey Sheridan, Board Certified Ophthalmologist. All authors agree to the contents of the final manuscript and case report.

Competing Interests
Author(s) disclose no potential conflicts of interest.

Disclosures and Ethics
As a requirement of publication author(s) have provided to the publisher signed confirmation of compliance with legal and ethical obligations including but not limited to the following: authorship and contributorship, conflicts of interest, privacy and confidentiality and (where applicable) protection of human and animal research subjects. The authors have read and confirmed their agreement with the ICMJE authorship and conflict of interest criteria. The authors have also confirmed that this article is unique and not under consideration or published in any other publication, and that they have permission from rights holders to reproduce any copyrighted material. Any disclosures are made in this section. The external blind peer reviewers report no conflicts of interest.

References
1. Traboulsi EI, Maumenee IH. Peters' anomaly and associated congenital malformations. *Archives of Ophthalmology*. 1992;110(12):1739–42. doi:1463415.
2. Hennekam RC, Van Schooneveld MJ, Ardinger HH, et al. The peters'-plus syndrome: Description of 16 patients and review of the literature. *Clin Dysmorphol*. 1993;2(4):283–300.
3. Ozeki H, Shirai S, Nozaki M, et al. Ocular and systemic features of peters' anomaly. *Graefes Arch Clin Exp Ophthalmol*. 2000;238(10):833–9.
4. Mirzayans F, Pearce WG, MacDonald IM, Walter MA. Mutation of the PAX6 gene in patients with autosomal dominant keratitis. *Am J Hum Genet*. 1995;57(3):539–48.
5. Weisschuh N, Wolf C, Wissinger B, Gramer E. A novel mutation in the FOXC1 gene in a family with axenfeld—rieger syndrome and Peters' Anomaly. *Clinical Genetics*. 2008;74(5):476–80. Retrieved from Google Scholar.
6. Shirai K, Okada Y, Nakamura Y, Saika S. Histopathological features in a case of peters' anomaly with acquired corneal staphyloma. *Case Rep Ophthalmol Med*. 2011;2011:418048. doi:10.1155/2011/418048
7. Bhandari R, Ferri S, Whittaker B, Liu M, Lazzaro DR. Peters anomaly: Review of the literature. *Cornea*. 2011;30(8):939–44. doi:10.1097/ICO.0b013e31820156a9.
8. Heath DH, Bruce Shields M. Glaucoma and peters' anomaly. *Graefe's Archive for Clinical and Experimental Ophthalmology*. 1991;229(3):277–280. Retrieved from Google Scholar.
9. Zaidman GW, Flanagan JK, Furey CC. Long-term visual prognosis in children after corneal transplant surgery for peters anomaly type I. *Am J Ophthalmol*. 2007;144(1):104–8. doi:10.1016/j.ajo.2007.03.058.
10. Bardakjian TM, Kwok S, Slavotinek AM, et al. Ocular genetics. *Development*. 2010;11(e10565):47. doi:10.1097/ICU.0b013e32834a3cdb.
11. Chang JW, Kim JH, Kim SJ, Yu YS. Long-term clinical course and visual outcome associated with Peters' Anomaly. *Eye*. 2012. doi:10.1038/eye.2012.128.
12. Ott EZ, Mills MD, Arango S, Getson AJ, Assaid CA, Adamsons IA. A randomized trial assessing Dorzolamide in patients with glaucoma who are younger than 6 years. *Archives of Ophthalmology*. 2005;123(9):1177–86. doi:10.1001/archopht.123.9.1177.
13. FDA Approved Drug Products. FDA approved drug products. Retrieved from http://www.accessdata.fda.gov/scripts/cder/drugsatfda/index.cfm. 2013.
14. Schaumberg DA, Moyes AL, Gomes JA, Dana MR. Corneal transplantation in young children with congenital hereditary endothelial dystrophy. Multicenter pediatric keratoplasty study. *Am J Ophthalmol*. 1999;127(4):373–8.
15. Cameron JA. Good visual result following early penetrating keratoplasty for peters' anomaly. *J Pediatr Ophthalmol Strabismus*. 1993;30(2):109–12.
16. Parmley VC, Stonecipher KG, Rowsey JJ. Peters' anomaly: A review of 26 penetrating keratoplasties in infants. *Ophthalmic Surg*. 1993;24(1):31–5.
17. Rao KV, Fernandes M, Gangopadhyay N, Vemuganti GK, Krishnaiah S, Sangwan VS. Outcome of penetrating keratoplasty for peters anomaly. *Cornea*, 2008;27(7):749–53. doi:10.1097/ICO.0b013e31816fe9a7.
18. Yang LL, Lambert SR, Lynn MJ, Stulting RD. Surgical management of glaucoma in infants and children with peters' anomaly: Long-term structural and functional outcome. *Ophthalmology*. 2004;111(1):112–7. doi:10.1016/j.ophtha.2003.02.002.
19. Gollamudi SR, Traboulsi EI, Chamon W, Stark WJ, Maumenee IH. Visual outcome after surgery for peters' anomaly. *Ophthalmic Genet*. 1994;15(1):31–5.
20. Basdekidou C, Dureau P, Edelson C, De Laage De Meux P, Caputo G. Should unilateral congenital corneal opacities in peters' anomaly be grafted? *European Journal of Ophthalmology*. 2011;21(6):695–9. doi:10.5301/EJO.2011.6317.