RETINAL DETACHMENT AND GIANT RETINAL TEARS IN ANIRIDIA

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SUMMARY

Clinical findings in aniridia may include corneal epitheliopathy, glaucoma, lens subluxation and cataract, and hypoplasia of the fovea and optic nerve. We report the occurrence of retinal detachment due to giant tears in four eyes of three children with aniridia. All eyes were buphthalmic and none had undergone lens or posterior segment surgery. All operated eyes underwent vitreolensectomy and silicone oil injection; useful vision was restored in two eyes. The pathogenesis and management of this previously unreported complication are discussed.

CASE REPORTS

Patient 1

Sporadic aniridia and buphthalmos were diagnosed at 6 weeks of age. At 4 months bilateral trabeculotomy was carried out with successful control of intraocular pressure. Bilateral lens subluxation was noted at 1 year, at which time corneal diameters were 13.75 mm, 12.5 mm and refraction was −6.25, −10.25 right and left respectively. A right convergent squint was present with manifest nystagmus. The best recorded visual acuities were 2/60 in each eye.

At 6 years of age, retinal detachment with a 360° giant retinal tear (GRT) was noted in the right eye with severe proliferative vitreoretinopathy (PVR). Following vitreolensectomy, silicone oil exchange, endolaser and retinal tacking, the retina was reattached but 1 month later redetached due to PVR and was deemed inoperable. No prophylaxis was performed on the left eye.

At 7 years of age, retinal detachment with a 300° GRT occurred in the left eye and was treated with vitreolensectomy, heavy liquid injection, silicone oil exchange and endolaser retinopexy. The retina remained flat and silicone oil removal was combined with epimacular membrane peeling 6 weeks later.

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At the age of 8 years the intraocular pressure was normal in both eyes. Early band keratopathy was noted in the left eye. The vision in the left eye was finger counting. Ocular axial lengths were 27.3 mm right, 25.8 mm left.

Patient 2

Autosomal dominant aniridia was diagnosed at birth and the left eye noted to be enlarged. At 5 months the left corneal diameter was 13 mm and left refractive error −7 dioptres. At 2 years subluxation of the left lens was noted. The best recorded visual acuities were 4/60 right and 3/60 left. Intraocular pressures were 18 mmHg right, 24 mmHg left with a cup:disc ratio of 0.6 in the left eye.

At 5 years a 120° GRT and retinal detachment were noted in the left eye. Vitreolensectomy with silicone oil and endolaser retinopexy produced retinal reattachment and the oil was removed 5 weeks after surgery.

At 6 years, the left vision was restored as 3/60 with marked anisometropia (left aphakia, right myopia). Extensive corneal pannus was noted at the most recent examination. The axial length of the left eye was 26.1 mm.

Patient 3

Autosomal dominant aniridia was diagnosed at birth. No surgery was undertaken. At 12 years of age the child presented to Moorfields with a history of recent deterioration of vision in the left eye. Nystagmus, corneal pannus and bilateral upward lens subluxation were noted. Intraocular pressure was 30 mmHg in the right eye. The left eye was found to have an inoperable total retinal detachment with a GRT and PVR.

DISCUSSION

Aniridia represents a spectrum of disorders with iris hypoplasia. It may occur sporadically or be inherited in an autosomal dominant fashion. The majority of

sporadic forms represents new autosomal dominant mutations. Some sporadic forms are associated with 11p13 deletion and Wilms' tumour and a variety of systemic syndromes exist.[1] The cornea typically shows progressive superficial vascular ingrowth from the limbus. Polar or cortical cataracts may develop, and the lens may subluxate, perhaps as a result of a molecular abnormality of zonule.[2] Glaucoma, although not present at birth, appears in childhood in up to 75% of cases. Optic nerve hypoplasia, foveal hypoplasia, refractive error, nystagmus and strabismus contribute to poor vision.

Neither retinal detachment nor giant retinal tears have previously been reported as complications of aniridia. Possible factors in the pathogenesis of retinal detachment in aniridia may include prior surgery, some vitreoretinal abnormality related to aniridia, and buphthalmic ocular enlargement.

No eye in this report had undergone lensectomy or surgery which involved vitreous manipulation, and it therefore seems unlikely that prior surgery was a factor in the genesis of detachment in these cases. Lens surgery in aniridic eyes may, however, predispose to retinal detachment; we are aware of two siblings with autosomal dominant aniridia, both of whom developed traction tears and retinal detachment following lensectomy.

Peripheral retinal abnormalities have been described in aniridia, and it is possible that such changes may be linked to detachment. Jesburg[3] noted the occurrence of multiple, small, circumferentially distributed white spots in the post-oral retina of 3 patients with aniridia; these had the staining attributes of lipid. These appearances were identified in all our cases (Fig. 1). White spots, however, are common in aniridia but retinal detachment is rare. In a histological case study of aniridia with gonadoblastoma and mental retardation, Anderson *et al.*[4] described an area of pathological vitreoretinal attachment in which small retinal strands entered the anterior vitreous. Vitreoretinal abnormalities were also noted by Seefelder[5] on histological examination of a patient with aniridia and buphthalmos. No such abnormalities were, however, noted in the eyes in this report. Finally, an abnormality of anterior vitreous might be inferred from the presence of lens subluxation in all our cases.

Myopia predisposes to retinal detachment, and in children may be associated with giant tears; in a series of juvenile detachments reported by Daniel *et al.*[6] giant tears occurred in 5 of 30 eyes with myopia unassociated with primary vitreoretinal degeneration. This predisposition is also associated with the ocular enlargement of buphthalmos. Of 19 buphthalmic eyes with detachments described by Cooling *et al.*[7] 2 had giant breaks and 2 giant dialyses. All the eyes we report were buphthalmic; corneal diameters were enlarged, phakic refractions were myopic, and axial lengths were increased. Ocular enlargement may thus predispose to retinal detachment in aniridia.

Giant retinal tears and ocular enlargement occur in children with the congenital megalophthalmos syndrome (CMS) as described by Scott.[8,9] CMS differs from the cases under study in that the myopia is congenital rather than acquired, characteristic vitreous abnormalities are present, and the incidence of detachment is far higher. Giant retinal tears may also arise in children with other congenital ocular abnormalities. Hovland *et al.*[10] described 8 cases of bilateral retinal detachment caused by giant retinal tears associated with nasal colobomas of lens and zonule; there were, however, no abnormalities of the uveal tract.

Symptomatic visual loss occurred in all cases reported here, and this symptom should suggest the need for examination of the posterior segment in aniridic patients. Photophobia, nystagmus, strabismus, corneal opacity, lens subluxation and cataract may, however, impair visualisation of the retina, and B-scan ultrasonography may be required.

Technical constraints on retinal reattachment surgery in buphthalmic eyes may be include poor visualisation of the peripheral retina due to corneal opacity, the presence of proliferative vitreoretino-

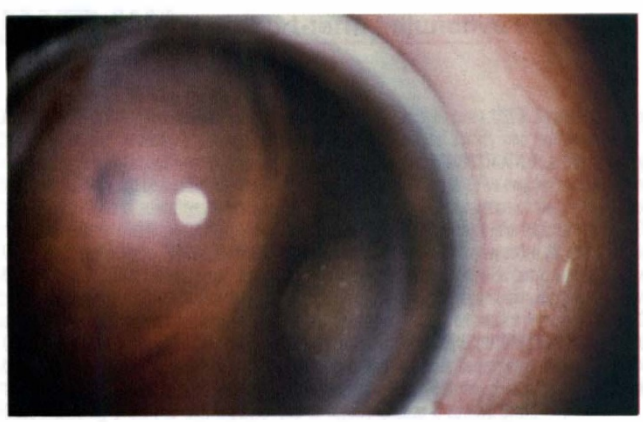

Fig. 1. *White spots in the pre-equatorial retina of an aniridic eye.*

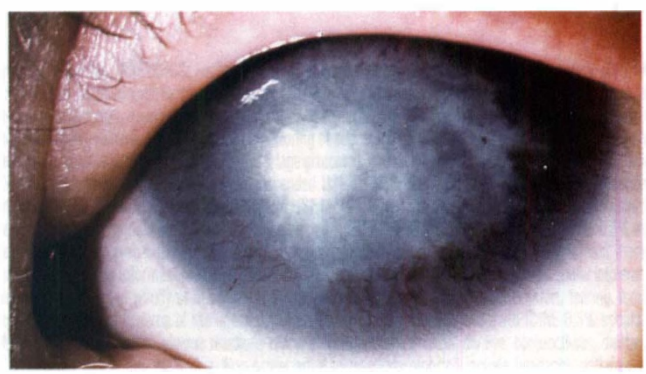

Fig. 2. *Keratopathy in an aphakic aniridic eye containing silicone oil.*

pathy and subretinal proliferations, and the need to maintain intraocular pressure during surgery at a level appropriate to both low scleral rigidity and compromised optic nerve perfusion.[11] Giant tears are associated with a high risk of proliferative vitreoretinopathy, and visualisation difficulties may be compounded by peripheral corneal pannus, lens opacity or lens subluxation. These considerations led to the use of vitreolensectomy with silicone oil injection in all cases in this report.

Hypotony or raised intraocular pressure may occur following retinal reattachment surgery in buphthalmic eyes.[11] In aphakic aniridic eyes, reformation of the anterior chamber following silicone oil injection poses particular problems and glaucoma may be exacerbated. Filtration surgery in this context is likely to carry a poor prognosis because of conjunctival scarring.[12] In the present series oil was removed from 2 eyes within 6 weeks of surgery, but was not removed from the eye in which surgery had been unsuccessful. Intraocular pressure control has, however, remained stable in all eyes to date.

Pre-existing corneal opacity associated with glaucoma, peripheral aniridic corneal pannus, silicone oil/endothelial contact,[13] post-operative corneal decompensation and band keratopathy may contribute to keratopathy following retinal reattachment surgery in aniridic eyes. In this series band keratopathy and more marked corneal pannus developed postoperatively in 2 eyes, and the most severe keratopathy (Fig. 2) occurred in the eye from which silicone oil was not removed, which argues for early removal of oil where indicated.

Reattachment of the retina with recovery of ambulatory vision was achieved in 2 of 3 operated eyes. In these young patients predetachment vision was poor and retinal detachment or other sight-threatening pathology uniformly bilateral. Vitreoretinal surgery in this context appears to offer a worthwhile prospect of retaining useful vision.

Key words: Aniridia, Buphthalmos, Giant retinal tear, Retinal detachment.

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