Fax: +44 191 2275246
E-mail: deepalivarma@hotmail.com

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Sir,
Buphthalmos in trisomy 13

Trisomy 13 is the rarest and most severe of the three viable autosomal trisomies, the others being 18 and 21. It is also the chromosomal abnormality most commonly associated with severe ocular defects, including microphthalmos, iris coloboma, and retinal dysplasia. The clinical picture of trisomy 13 was described in 1656 by Thomas Bartholin, and in 1960, Klaus Patau¹ reported the finding of an extra D chromosome in such a patient, now known to be an additional chromosome 13. The typical dysmorphic features include microcephaly, hypertelorism, micrognathia, low-set ears, cutis aplasia, cleft lip and palate, and polydactyly. Internal structural defects affecting the heart and brain are common, with high mortality in the neonatal period. Survivors exhibit severe developmental delay.

Case report

The neonate we describe was a female born prematurely by caesarean section at 28 weeks gestational age. The mother, age 39 years, had pre-eclampsia and the foetus was growth retarded, weighing just 630 g at birth. She was noted to have micrognathia and microstomia, low-set ears, bifid uvula, wide scalp defect between anterior and posterior fontanelles, and bilateral talipes. Ocular examination showed hypertelorism. The globes were buphthalmic and bilateral corneal stromal oedema was present, obscuring fundal detail. Examination with the indirect ophthalmoscope revealed aniridia and some evidence of an iris root only. Intraocular pressures by Perkins applanation tonometry were 30 mmHg OD and 40 mmHg OS. Treatment with topical dorzolamide was commenced; however, before the eyes could be re-evaluated, a diagnosis of Patau's syndrome was confirmed cytogenetically, and in view of severe respiratory difficulty all treatment was withdrawn.

Comment

In his original description of a patient with trisomy 13, Patau states 'no organised ocular tissue could be seen or palpated'. Case series detailing the ocular characteristics of trisomy 13 have since been published.^{2–4} The most

frequent findings are microphthalmia, coloboma of the iris and ciliary body, and retinal dysplasia. Other abnormalities described include PPHV, cataract, cyclopia, and primary aphakia. We have found four previously reported cases of congenital glaucoma associated with trisomy 13.

The first such description is by Keith⁴ in a review of ocular manifestations and histopathology in 10 cases of trisomy 13. He noted bilateral buphthalmos in one case, gestational age not stated. No iris defect is described. Hinzpeter *et al*⁵ reported a trisomy 13 neonate born at 35 weeks gestational age with corneal diameters of 15 × 15 mm bilaterally, and IOPs of 33 mmHg OD and 50 mmHg OS. Histological examination demonstrated anterior inserted iris root but normal iris leaf. Lichter and Schmickel⁶ described a trisomic infant born at term, who developed corneal enlargement and clouding at 4 months of age. IOPs were 59 and 40 mmHg. The iris appeared normal. In 1981, Reardon *et al*⁷ reported an unusual trisomy 13 mosaic, the predominant cell line being trisomic for the distal portion of the long arm of chromosome 13. Born at 37 weeks gestation, ocular examination showed left microphthalmos and iris coloboma. Glaucoma of the right eye was recognised at 1 year with buphthalmos. The right iris was atrophic with stromal defects inferiorly. IOP was 50 mmHg OD, 15 mmHg OS.

The case of trisomy 13 we have described is notable both for established buphthalmos at just 28 weeks gestational age, and the lack of iris. The early delivery of this infant tells us that buphthalmos can take place as early as the second trimester, contrasting with presentation at 1 year of age in the case of the trisomy 13 mosaic. Mosaicism was excluded in our case on the basis of a 10 cell karyotype, coupled with the severity of the phenotype.

The second unusual finding, the almost complete lack of iris tissue, has not been described in the literature in association with trisomy 13, nor is it a feature of the above cases of glaucoma in trisomy 13. Iris coloboma, however, is a typical feature and while we have not excluded specific secondary chromosomal defects, a severe colobomatous-type defect would seem the most likely aetiology.

A normal chromosome 13 complement seems to be essential for the proper development of the eye, and while trisomy 13 often results in a microphthalmic eye at birth, it can exceptionally give rise to buphthalmos. Hoepner and Yanoff⁸ emphasised a characteristic 'dysembryogenesis' of the anterior segment in a histological review of trisomy 13 patients, finding microphthalmic eyes with immature drainage angles. We speculate that if the drainage structures are disproportionately underdeveloped compared with the

ciliary body, then congenital glaucoma may arise. A genetic basis for many developmental glaucomas is increasingly being demonstrated, and trisomy 13 is a further addition to the list.

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R Bunting and J Leitch

HM Stanley Hospital, UK Sutton Eye Unit, UK

Correspondence: R Bunting
Tel: +44 7906711844
Fax: +44 208 8770 2896
E-mail: rmbunting@doctors.org.uk

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Sir,
Retinal macroaneurysm with macular hole and subretinal neovascular membrane

A rare case of an untreated retinal macroaneurysm with an underlying subretinal neovascular membrane and a macular hole is presented.

Case report

A 71-year-old hypertensive lady presented to the ophthalmic casualty department with visual distortion

and sudden onset of floaters in her right eye. There is no history of headache, ocular pain, flashing lights, or trauma. She was a known case of left branch retinal artery occlusion. Her best-corrected Snellen acuity was 6/5 in the right and 6/9 in the left eye. Anterior segment examination was normal. Fundus examination revealed a macroaneurysm involving the superotemporal retinal artery in the right eye with a large subretinal, preretinal haemorrhage, and a mild vitreous haemorrhage. The patient was advised about the diagnosis, a carotid Doppler requested and a follow-up appointment arranged in 8 weeks.

On review at 8 weeks, a significant reduction in vitreous haemorrhage and a thrombosed macroaneurysm in the supratemporal region with accompanying retinal and macular oedema was recorded. Laser treatment was deferred due to the presence of a large subretinal haemorrhage. Doppler studies of internal carotids showed 50% stenosis in the left carotid with no significant disease on the right side. She was on aspirin and was advised a second antiplatelet agent.

On a further review in 6 weeks, her visual acuity was 6/60 in right eye and 6/9 in left eye. Retinal examination still showed an occluded macroaneurysm but now with an underlying subretinal neovascular membrane and a macular hole (Figure 1), the presence of which were confirmed on fundus fluorescein angiography (Figure 2), indocyanine green angiography, and ocular coherence tomography (Figure 3). In view of limited visual benefit from laser treatment, the position and size of the subretinal neovascular complex, no treatment has been planned.

Figure 1 Colour fundus photograph demonstrating the macroaneurysm (a), choroidal neovascular complex (b), and a macular hole (c).