CORRESPONDENCE

The south Asian cataract management study

EDIToR,—In reading the preliminary report evaluating the clinical outcome of intracapsular cataract extraction (ICCE) with aphakic correction either via anterior chamber intraocular lens (AC IOL) implants or aphakic spectacles in a developing country setting I was given cause for alarm with regard to the assessment of the differences in visual outcome.

While the study is ongoing, I think it is important to recognise that a varying percentage (16–25%) of standard ICCE patients in a developing country setting will lose their spectacles and have problems adapting to the magnified image, forcing some patients not to wear them at all.2,4 By the same token, the quality of vision and life enjoyed by those patients with AC IOL implants is likely to be superior to those patients relying on aphakic spectacles, or none at all. However, the report’s stress on the stringent use of visual acuity measures alone and its conclusion that ‘There was no significant difference in visual outcome between study groups’ fails to address adequately the variation attributable to spectacle and non-spectacle using aphakic patients in determining the overall differences in visual outcomes between the two study groups.1 In this regard, recent instruments developed to measure both the preoperative and postoperative visual function and quality of life aspects of cataract patients within the context of a developing country setting offer an avenue for further exploration of the relation between visual acuity and overall patient satisfaction with the visual outcome of cataract surgery.7

PETERS’ ANOMALY

EDIToR,—I read with interest the recent letter by Saitoh et al describing an infant with both Peters’ anomaly and Axenfeld-Rieger anomaly.8 Unfortunately the discussion which follows the case description is marred by a failure to mention some significant discoveries which have been made in this field over the past few years. I would like to bring to their attention the finding of a deletion of the Pax6 gene in a child with aniridia and Peters’ anomaly, and an intragenic mutation of the same gene in a family with heterogeneous anterior segment malformations including Peters’ anomaly.2,3 Deletions and mutations of the Pax6 gene were sought in these patients for the following reasons. The Pax6 gene was known to be deleted or mutated in aniridia, and cases of aniridia with Peters’ anomaly had been described.1,4 Furthermore, deletions and mutations of Pax-6, the mouse homologue of Pax6, had been found in the Small eye mouse which phenotypically resembles aniridia and Peters’ anomaly.5,6

The Pax6 gene plays a crucial role in development as illustrated by the discovery of Pax6 gene homologues in Caenorhabditis elegans, Drosophila, Ciona intestinalis elegantis, and sea urchin. Recent experiments using targeted expression in Drosophila led to the production of supernumerary ectopic eyes giving support to the idea that Pax6 is the ‘master control’ gene for eye formation.7

The expression pattern of Pax6 has been investigated in both human and mouse.11 These studies found expression in ocular precursor derivatives from neuroectoderm and head surface ectoderm which give rise to the corneal epithelium, the lens, the posterior layers of the iris, the ciliary body, the retina, and the optic nerve. The primary defect in aniridia and Peters’ anomaly caused by Pax6 mutations must therefore lie within these tissues and fits well with the spectrum of clinical abnormality seen in these disorders—that is, corneal opacification at the subepithelial level, incomplete separation of the lens, persistent lens stalk, central corneal leucoma, iridocorneal adhesions, iridocorneal adhesions, iris hypoplasia, cataract, macula dysplasia, and retinopathy. In lines of mice with targeted mutation of midbrain neural crest cells has been found in the Uchida rat which has an intragenic mutation of the rat homologue of Pax6.4,5 However, this does not affect the arrival of neural crest cells at the eye and since the Pax6 gene is not known to be expressed in this tissue it is unlikely to be the site of primary pathology.

Although the concurrence of Peters’ anomaly and Axenfeld’s anomaly is an interesting finding it may be a little premature to conclude that this is the usual case and that hitherto remained undetected. I would like to suggest that an alternative explanation is that due to a single gene causing a range of phenotypes with the primary abnormality residing within the expressing tissue—that is, neuroectoderm and head ectoderm, and secondary abnormalities occurring within tissues with which it interacts—that is, neural crest cell mesenchyme. This principle should still hold true for cases of Peters’ anomaly not due to abnormality of the Pax6 gene. Some of these may result from a primary disorder of neural crest cells but this has yet to be proved.

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11 Wilder H, Gruss P. Pax-6, a murine paired box gene, is expressed in the developing CNS. Development 1991;113:1435–49.

Reply

EDIToR,—We also have an interest in the recent discoveries of the association between anterior segment anomalies and Pax6 locus. At present, we are analysing the Pax6 locus of an appropriate congenital cataract patient—single strand conformation polymorphism, and sequencing analysis methods.14 Jordan’s alternative explanation is interesting,