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Anterior polar cataract: clinical spectrum and genetic linkage in a single family

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Abstract

Anterior polar cataract can occur as a sporadic finding, in association with other ocular abnormalities or as an inherited, autosomal dominant disorder. We have demonstrated linkage in a family with autosomal dominant anterior polar cataract to the short arm of chromosome 17, locating the gene to the region 17p12–13. All affected members of this large family had an opacity at the anterior pole of the lens that varied only in size and the effect on visual acuity. Anterior polar cataract is thought to have a minimal effect on visual acuity although in the affected members of this family there was a high incidence of unilateral amblyopia.

Key words Anterior polar, Cataract, Autosomal dominant, Linkage, Phenotype

Inherited cataract is a significant cause of visual impairment in childhood and is most often transmitted as a congenital autosomal dominant (AD) trait.[1] Eight different loci have now been mapped for AD cataract[2–9] but the underlying genetic abnormality is known in only one case: the Coppock-like cataract has been shown to be related to the overexpression of a γ-crystallin pseudogene.[10] We have demonstrated linkage of an AD anterior polar cataract (APC) to the short arm of chromosome 17[11] in a single family and in this paper describe the clinical spectrum of the disease.

Patients and methods

The patients were from a large family with AD anterior polar cataract (Fig. 1) identified from the genetic data base at Moorfields Eye Hospital. Family members were invited to attend the hospital for an eye examination. The lens was examined by slit lamp and anterior segment photography was taken where possible. Blood was taken for DNA extraction to be used for the linkage analysis.

Results

The results of the molecular genetic study demonstrating linkage of the anterior polar cataract in this family to chromosome 17p have already been published.[11] Anterior polar cataracts were seen in all the affected family members. Thirty family members were examined and 19 found to be affected. Seven patients had undergone surgery and the phenotype was examined in the remaining 12 cases. The opacity was always in the same position of the lens, the anterior pole, but varied in severity. The larger opacities were pyramidal with a base of 4 mm and extended into the anterior chamber (Figs. 2, 3), whilst the smaller opacities consisted of a flat plaque measuring less than 1 mm in diameter (Figs. 4, 5). In one case the only sign of an anterior pole abnormality was an irregularity of the anterior capsule (Fig. 6). Seven of the affected patients underwent surgery, all between the age of 1 and 6 years, with a final visual acuity of 6/18 to NPL. Two patients who had a visual acuity of NPL had both developed glaucoma and retinal detachment. One affected patient who had not undergone surgery but had minimal opacities had a visual acuity of 6/6 in both eyes.

Ten of the 12 affected patients who had not undergone surgery were amblyopic in one eye with a visual acuity of 6/12 to 6/36 in the worse eye. The amblyopic eye was always the eye with the larger opacity.

Discussion

Anterior polar cataract has been reported to have a minimal effect on visual acuity.[12,13] In this family unilateral amblyopia was common, suggesting that children with anterior polar cataracts should be offered regular follow-up.[14] The unoperated patients had a better visual outcome than those who had surgery for their anterior polar cataracts. Although the visual outcome may reflect the severity of the cataract it suggests that better long-term results may be achieved with conservative treatment.

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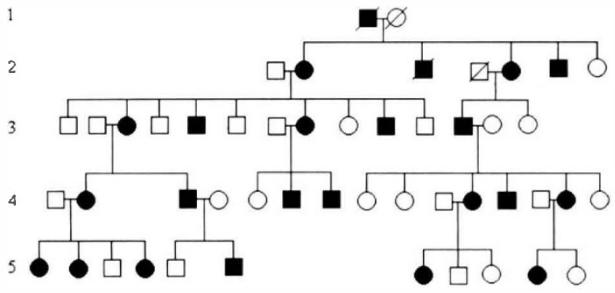

Fig. 1. *Pedigree affected by anterior polar cataract.*

AD cataract may show considerable phenotypic variability between family members.[15] In this family, however, all affected individuals had lens opacities confined to the anterior pole of the lens. We have demonstrated that the anterior polar cataract in this family is linked to the short arm of chromosome 17.[11] The mechanism by which the lens opacity is confined to the anterior pole is unclear but identification of the precise genetic mutation will help in understanding the pathogenesis. There are at present no candidate genes in this region on chromosome 17p, but research to identify the causative gene continues.

Fig. 2. *Anterior polar cataract. Slit lamp photograph of a pyramidal cataract from individual 4:2.*

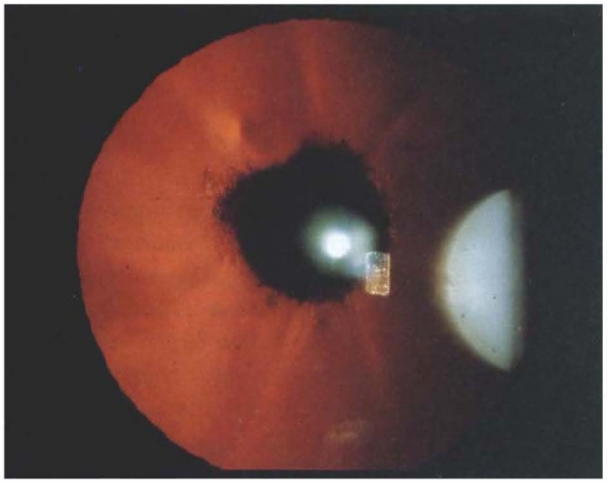

Fig. 3. *Anterior polar cataract. Retroillumination of the pyramidal cataract from individual 4:2.*

Fig. 4. *Anterior polar cataract. Slit lamp photograph of a plaque-like opacity from individual 4:3.*

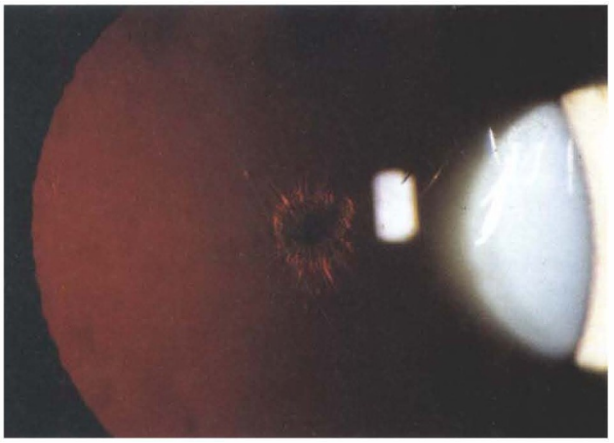

Fig. 5. *Anterior polar cataract. Retroillumination of the plaque-like opacity from individual 4:3.*

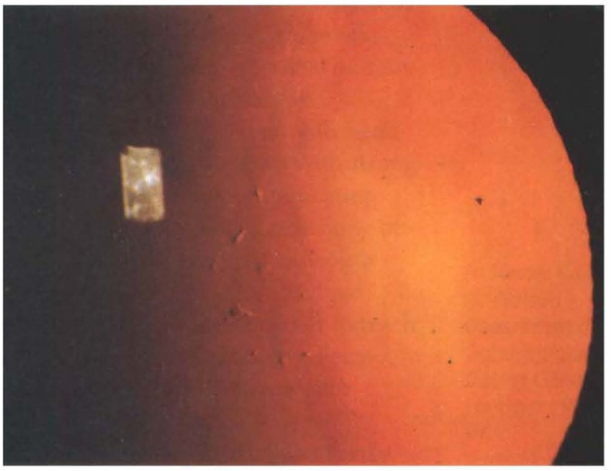

Fig. 6. *Anterior polar cataract. Retroillumination of an anterior capsular irregularity from individual 5:6.*

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