# Congenital aphakia in Peters' anomaly syndrome

## A Case Report

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ABSTRACT.

**The authors report a case of congenital bilateral corneal opacities in which one of the eyes was enucleated because of malignant glaucoma and corneal perforation. Corneal defects and iridocorneal adhesion were found, but aphakia was the major pathologic ocular finding. The clinical picture and pathology study indicated this case as a Peters' anomaly presenting congenital aphakia.**

**Key words:** Peters' anomaly - glaucoma - monolateral aphakia - cornea.

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Peters' anomaly is a congenital disease that affects mainly the posterior cornea (Alkemade 1969). It is most often bilateral and characterized by localized defects in the endothelium and Descemet's membrane resulting in corneal clouding (Naganishi & Brown 1971). Iris and/or lens adhesion to the cornea are described together with absence of endothelium and Descemet's membrane (Stone et al. 1976).

To our knowledge congenital aphakia associated with Peters' anomaly has only been reported in one case affected unilaterally, described by Harris et al. (1980). We describe the clinical findings of a 6-day-old newborn observed in our department, affected by bilateral corneal opacities, malignant glaucoma and unilateral aphakia.

The authors also report the histopathological aspects of the eye that underwent enucleation due to refractory increased IOP and corneal perforation.

## Materials and Methods

A 6-day-old newborn boy was examined in our department for bilateral corneal leucoma. He underwent examination under anaesthesia (EUA). Slit-lamp biomicroscopy, applanation tonometry and ocular B-scan ultrasonography were performed. Retinal and optic nerve functions were evaluated by means of ERG and VEP. A complete pediatric work-up was carried out, including karyotype examination. Six days after the first examination the right eye was enucleated and examined histopathologically.

## Results

Clinical inspection showed exophthalmos of the right eye.

Slit-lamp examination revealed bilateral vascularized leucoma associated with a central corneal ectasia of the right eye. The corneal diameter was 24 mm RE and 11 mm LE. IOP was 33 mmHg RE and 28 mmHg LE. Fundus examination was not possible due to corneal opacities. Ultrasonography showed an axial length of 24.5 mm RE and 16.5 mm LE. In the RE B-scan ultrasonography revealed an enlargement of the globe and aphakia. No abnormal echos were detected in the vitreal cavity and the retina appeared to be attached. In the LE ultrasonography was within normal limits. Photopic ERG was extinguished in the RE and extremely reduced in amplitude in the LE. Flash VEP was not obtainable in either eye. No anomalies were detected by the pediatric work-up. Routine hematologic exam was within normal limits including the TORCH complex, VDRL and TPHA. The karyotype was 46XY.

Macroscopic examination of the enucleated right eye revealed abnormal anterior segment structures which included: corneal leukoma, iris and ciliary body atrophy. The lens was not detected (Fig. 1).

A histopathological study showed that corneal thickness was extremely enlarged. The collagen bundles of the stroma were swollen and densely stained. In the central portion of the cornea the Descemet's membrane and endothelium were interrupted. The iris and ciliary body were atrophic (Fig. 2). The iris was adherent to the posterior surface of the cornea and the ciliary bodies were turned forward. The sclera, choroid and retina were thinned.

A deep excavation of the optic disk was observed (Fig. 3).

## Discussion

This paper describes a case of congenital bilateral corneal opacities, iridocorneal adhesion and defects associated with monolateral aphakia. The characteristics of the anterior segment of the eye in Peters' anomaly vary significantly, but they commonly include glaucoma, which may be congenital, infantile or juvenile, and dense central opacities, less commonly, spontaneous corneal perforation (Varley et al. 1987) and retinal detachment (Traboulsi & Maumenee 1992). We believe that the findings observed in our case are consistent with those of a Peters' anom-

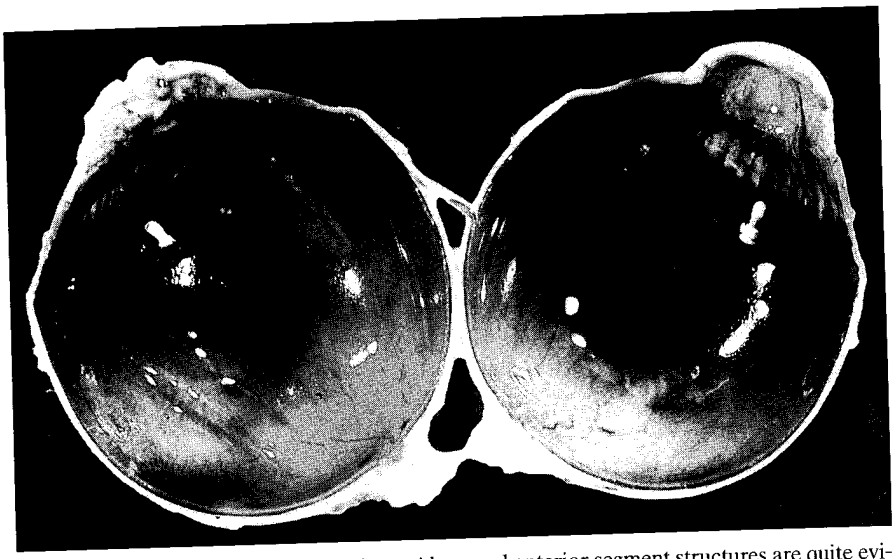

**Fig. 1.** Macroscopic view of sectioned eye. Abnormal anterior segment structures are quite evident: corneal leukoma, iris and ciliary body atrophy, ectasia of the sclera and, finally, aphakia.

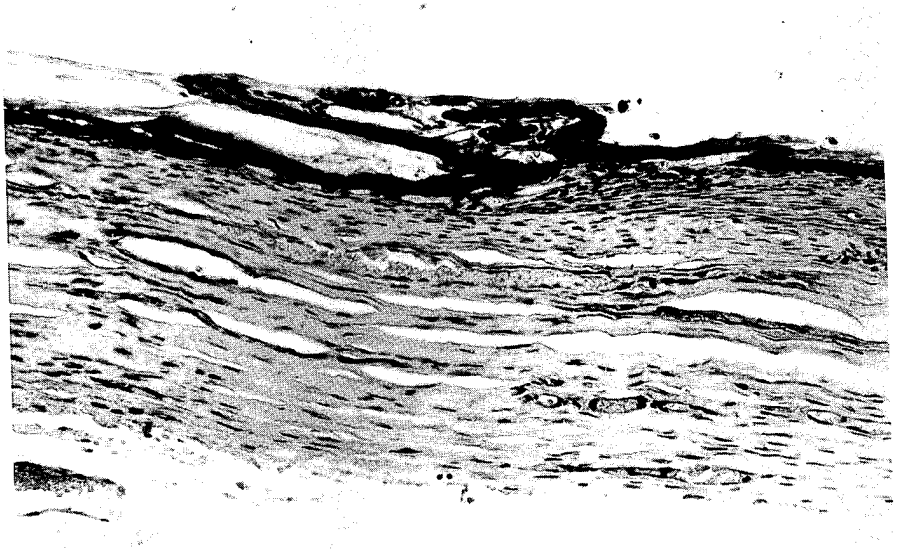

**Fig. 2.** Iris and ciliary body atrophy was confirmed by the histopathology.

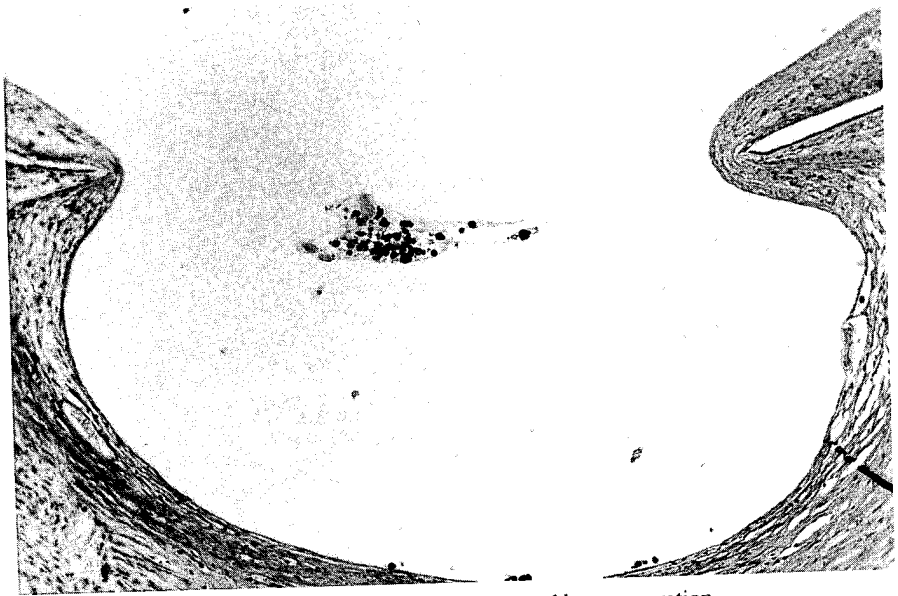

**Fig. 3.** Histologic section of optic disk revealed a deep and large excavation.

aly, although the characteristics observed have some modifications from those usually described in this condition, particularly related to the presence of a high pressure glaucoma. In addition to the expected findings we were able to observe also a monolateral congenital aphakia (Harris et al. 1980). Congenital aphakia may be either primary or secondary (Manschott 1963; Duke-Elder 1963). Usually, the former occurs associated with severe deformities of the globe which include anterior segment abnormal formation. Secondary aphakia is characterized by degeneration or absorption of the lens after it has developed, but usually some lens remnants are left. Moreover, eyes with secondary aphakia are less deformed than those with primary aphakia. Our patient's globe had multiple anomalies, no remnants of the lens, and thus we believe our case is an example of primary aphakia. The histopathological examination confirmed the clinical characteristics, disclosing also Descemet's membrane and endothelial defects. Our histopathologic report is consistent with a primary defect of the ectoderm with neural crest anomalies justifing disorganized corneal stroma and anomalies of the angle. According to recent reports (Tripathi & Tripathi 1989; Nucci & Manitto 1995), a defect of the neural crest cells is responsible for the genesis of some anterior chamber structures. A particular histopathological aspect was the scleral and chorioretinal thinning associated with deep excavation of the optic disk. Peters' anomaly usually includes normal or reduced volume of the eye (Bateman et al. 1984), but we relate the increased axial length and anatomical features of our case to increased IOP (Koster & van Balen 1985).

A number of chromosomal abnormalities are described in patients with Peters' anomaly (Traboulsi & Maumenee 1992; Bateman et al. 1984; Cibis et al. 1985). Our case does not reveal abnormalities of the karyotype, in accord with Traboulsi & Maumenee (1992) who reported that no single chromosomal abnormality has been consistently associated with Peters' anomaly. However, a point mutation of the gene responsible for the anterior segment development or of a homeotic gene (Kessel & Gruss 1990) controlling the development of the eye cannot be excluded.

This case of monolateral aphakia in Peters' anomaly seems to confirm that in Peters' syndrome a large number of different defects may be present. The last aspect has been described in patients with a

mutation or a deletion of a homeotic gene that controls the differentiation of primordial cells and the development of different body segments (Hanson et al. 1994).

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