Retinal pigment epithelial hamartoma—unusual manifestations

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SUMMARY Hamartoma of the retinal pigment epithelium is an uncommon tumour of young adults. We have seen 2 patients with this clinical diagnosis, both with unusual manifestations. In one patient growth of the tumour was observed over a 5-year period. In the second patient arterial-arterial anastomoses were detected at a site distal to the tumour.

Retinal pigment epithelial (RPE) hamartomas are rare tumours that are classically described as having no growth potential.¹ They have on occasion been misdiagnosed as juxtapapillary choroidal melanomas, especially in the small percentage of RPE hamartomas that were believed to show growth.² We have recently seen an adolescent with an RPE hamartoma showing photographic and angiographic evidence of marked growth. We also present a patient with an RPE hamartoma associated with arterial-arterial anastomoses in a site distal to the tumour.

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Case reports

CASE 1

This 12-year-old white male was first examined on 31 October 1977. He had been referred for evaluation of strabismus. His best corrected visual acuity was RE 20/200, LE 20/20. An exotropia of 20 D was present. The anterior segment was normal. Tension by applanation tonometry was 18 mmHg OU. The left fundus was normal. The right optic disc was partially obscured by a gliotic mass extending superiorly (Fig. 1A). The retinal vasculature in the area of the mass was markedly tortuous. There were some fine striae

Figs. 1B and 1C Fluorescein angiography. B: Venous phase, showing vascular tortuosity.

Fig. 1C Late phase, showing leaking vasculature and staining of glial tissue.

Figs. 1A-1C A 12-year-old white male with an exotropia of 20 D and a visual acuity of 20/200 OD, 20/20 OS. 1A: There is marked vascular tortuosity overlying the optic disc. Gliotic tissue is noted along the superior part of the disc, extending superiorly.

Fig. 3A A 16-year-old white female with visual acuity of finger counting OD, 20/20 OS. The nasal border of the tumour shows a scalloped, hyperpigmented border blending into the adjacent normal retinal pigment epithelium. The origin of the inferotemporal artery is difficult to discern (arrow).

Fig. 1D Five years later. VA 20/400 OD, 20/20 OS. There is increased vascular tortuosity with a marked increase in the size of the tumour. Note the flat, pigmented, feathery border of the tumour as it blends into the retinal pigment epithelium (arrow). There is also preretinal haemorrhage along the superonasal aspect of the disc and obscuration of the temporal portion of the disc.

Fig. 3D Fundus photograph shows a second area of arterial-arterial anastomosis temporal to the macula.

in the retina, heading out toward the foveal region. Fluorescein angiography demonstrated the retinal vascular tortuosity, with late leakage into the tumour mass (Figs. 1B, 1C). Our impression was an RPE hamartoma. Observation was suggested.

He was seen again over 5 years later on 14 December 1982. His vision in the right eye had decreased to 20/400. Fundus examination revealed a

striking increase in size in the ophthalmoscopic appearance of the mass (Fig. 1D). There was an increase in the gliotic portion of the mass with increased retinal vascular tortuosity. Small vessels within the tumour mass were discernible. The flat pigmented border of the tumour blended into the surrounding RPE (arrow). Fluorescein angiography demonstrated the tumour vasculature with late staining and showed graphic evidence of increased size (Figs. 1E, 1F). Ultrasonography performed at the time of this examination revealed a mildly elevated mass anterior to the optic nerve head with high internal acoustical interfaces (Figs. 2A, 2B).

Figs. 1E and 1F Fluorescein angiography of patient in Fig. 1D. E: Venous phase, showing marked vascular tortuosity involving an area 4×4 disc diameters in size. The perifoveal capillary bed is distorted.

CASE 2

A 16-year-old white female was examined on 15 October 1982. She had a history of strabismic amblyopia since the age of 5. She had been told of a vascular anomaly in the left eye several years previously. Vision was RE 20/20, LE 20/40. Examination of the anterior segment was normal. A 10 D left esotropia was present. The right fundus was normal. The left fundus revealed a minimally elevated gliotic mass extending on to the optic disc, and tortuosity of the retinal vasculature consistent with a clinical diagnosis of RPE hamartoma (Fig. 3A). There was no clearly defined inferotemporal artery (arrow), and on fluorescein angiography the inferior arterial circu-

Fig. 1F Late phase, showing leakage throughout the tumour and a disc haemorrhage causing a blocking defect.

Figs. 2A and 2B Ultrasonography. A: B-scan: mildly elevated mass anterior to the optic nerve head.

lation was seen to fill, with a slight delay, by arterial-arterial anastomoses which passed through the macula (Figs. 3B, 3C). A second site of arterial-arterial anastomosis was detected temporal to the fovea (Fig. 3D).

Discussion

A retinal pigment epithelial hamartoma is an uncommon tumour occurring in the first and second decades of life. It is most frequently located in the juxtapapillary region; however, a sizable number of eccentrically located tumours have been reported in all parts of the retina.³ Affected patients may be asymptomatic until late childhood, or they may notice blurred vision, metamorphopsia, distortion, or a negative scotoma. A high percentage of affected individuals have strabismus or have been diagnosed

Fig. 2B A-scan: high internal acoustical interfaces.

Fig. 3B Fluorescein angiography of arterial phase shows marked delay in filling of the inferior retinal vasculature.

as amblyopic. The lesion may be compatible with good visual acuity, with many patients seeing 20/70 or better. Visual acuity may vary, as contraction of glial tissue within the tumour causes increased traction on the macula. There is no involvement of the anterior segment, and no associated systemic disease. There is one reported case showing late development of peripapillary subretinal neovascularisation.⁴

RPE hamartomas generally show no growth potential. Cardell and Starbuck² reported a case of an RPE hamartoma which was enucleated on the basis of suspected clinical growth over a period of 4 months. Photographs of the lesion were not reported. They concluded that hamartomas may grow during periods of bodily growth as a whole, but do not show progressive continued growth. Laqua and Wessing⁵ described 6 patients with a diagnosis of RPE hamartoma. In one 16-year-old male they thought the tumour might be growing but suspected that this may have been just increasing surface contracture. The first patient we described showed documented growth of this tumour over a 5-year period during puberty.

The retinal vasculature not involved by the tumour is always reported as normal. However, we present a patient with an RPE hamartoma and 2 separate areas of arterial-arterial anastomoses. In one of these areas

Fig. 3C Fluorescein angiography shows arterial-arterial anastomosis, which is going through the macula to supply the inferior retinal arterial circulation.

this collateral circuit appears to supply the inferior half of the retina with arterial blood. The anastomoses may be either congenital or acquired. In the former case the tumour could have interfered with normal vascular development, while in the latter case tumour growth could have caused inferotemporal arterial obstruction. These findings show that an RPE hamartoma could have affected the retinal vasculature distal to the tumour. We do not believe this has been reported previously.

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