Sir,

Pigmented paravenous retinochoroidal atrophy: evidence of progression to macular involvement in a family with a 42-year history

Pigmented paravenous retinochoroidal atrophy (PPRA) is a rare pigmentary retinopathy that remains poorly understood. Patients with this condition show a striking pattern of sharply outlined zones of atrophy of the retinal pigment epithelium that follow the course of the major retinal veins. These may extend posteriorly and be confluent with zones of atrophy surrounding optic disc. The condition is bilateral and may commence in childhood. In this paper we report a rare case of macular involvement by the disease and the unusual association with a milder picture in the patient's mother. We have unique follow-up of 42 years with significant deterioration of vision due to progression of the disease to partial sight registration.

Case reports

Case 1. A white male initially presented as a child in 1957 aged 18 months with left convergent squint. In 1963 fundal examination recorded 'dark grey patch at macula and fine pigmentary changes in lower half of left fundus, normal right fundus'. Corrected vision at this time was 6/6 right eye and 2/60 left eye. In 1971 he was reviewed and, whilst visual acuity was maintained at the same level, both maculae showed 'fine pigmentary changes', as well as paravenous atrophy (Fig. 1a). In 1990 (aged 34 years) his visual acuity had dropped to 6/12 right eye and CF left eye: his pigmentary changes had progressed and colour vision was 4/15 on Ishihara plates. The electro-oculogram (EOG) and scotopic electroretinogram (ERG) were normal, but photopic ERG was reduced in the left eye, normal in the right. Visual fields showed a right central scotoma.

In 1996 the patient had noticed increasing problems with central vision, which was reduced to 6/24 right eye and CF left eye. His vision was worse in bright light with normal night vision. Fundal photographs showed definite progression as demonstrated in Fig. 1b. EOG was now bilaterally reduced, ERG was slightly reduced in the left eye and the right visual field scotoma had progressed. Fluorescein angiography showed extensive areas of hyperfluorescence, especially gathered around the major retinal veins and in the macular area corresponding to areas of atrophy of the retinal pigment epithelium (Fig. 2). Changes in the angiogram are more dramatic than the colour photograph, suggesting involvement of the choriocapillaris.

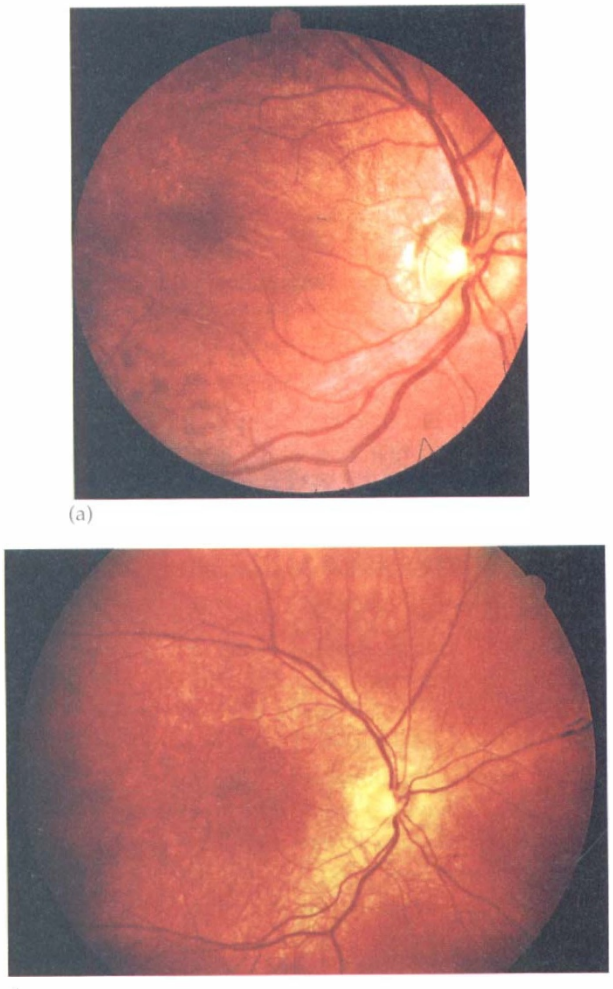

Fig. 1. *Case 1. (a) Colour fundus photograph of the right eye showing fine pigmentary changes. (b) Colour fundus photograph of the right eye showing macular and paravenous atrophy.*

Case 2. A white woman presented in 1964 aged 38 years with reduced vision of the right eye associated with right visual field constriction. Her vision was recorded as 6/9 right and 6/5 left. There was no family history of eye disease apart from her son (case 1). Fundal examination at this time showed 'fine pigmentary changes along vessels in right eye with similar but less obvious changes in left fundus'. A diagnosis of atypical retinitis pigmentosa was made. In 1977, aged 51 years, she presented with angle closure glaucoma in the right eye with vision of NPL right eye and 6/9 left eye. The left fundal appearance was described as 'retinitis pigmentosa with peripapillary atrophy' and visual fields showed mid-peripheral field loss. At her last clinic visit in 1996 she had a fundal appearance as shown in Fig. 3 with a visual acuity of 6/9. ERG was absent in the blind right eye and present but of slightly reduced amplitude in both photopic and scotopic testing in the left eye. Even at this late stage there was no difficulty

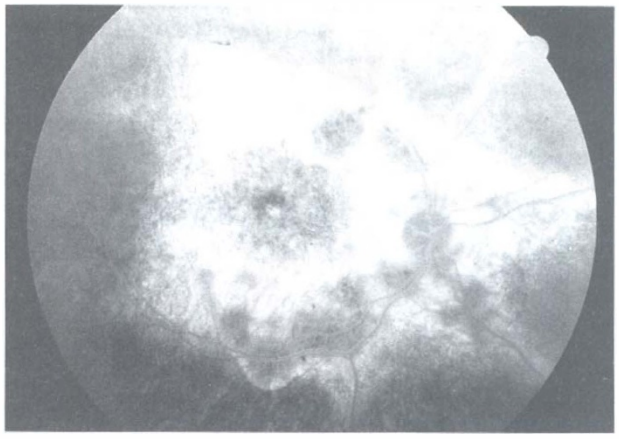

Fig. 2. *Case 1. Fundus fluorescein angiography of the right eye demonstrating extensive hyperfluorescence around major retinal veins and the macular area.*

with night vision. She died in 1998 of multiple myeloma. There are no further members of the family alive for examination.

Comment

Brown first described PPRA as 'retino-choroiditis radiata' in 1937[1] and the first use of the term pigmentary paravenous choroidoretinal degeneration is in a review by Franchescetti[2] in 1962. Usually it is described as sporadic and non-progressive. There are very few hereditary cases in the literature.[3–5] Pearlman *et al.*[6] and Noble and Carr[7] have reported mild progression in two 'sporadic' cases. Macular involvement has been reported only rarely[4,5,8] and ours (Case 1) is the first case with documented progression of macular involvement – seen photographically, on visual fields and on electrophysiology.

There has been a great variety of reported electrophysiological findings, from normal to reduced ERG and EOG.[3,4,6,7] This variation in findings may well be because of the varying ages of patients and severity of disease, or alternatively may mean that several conditions can present in this way. Interestingly, early onset angle closure glaucoma,[5] hypermetropia and esotropia[4,5] have been reported in other families with this condition. There is known to be variable expressivity even within one family.[4] This may mean that 'sporadic' cases are familial if asymptomatic relatives are carefully examined. Inheritance could be autosomal dominant or X-linked. PPRA can mimic retinitis pigmentosa but can be distinguished from it by the absence of night blindness, minimal ERG abnormalities and typical appearance. Other differential diagnoses include peripapillary choroidal atrophy, radiating peripapillary pigmentary degeneration and helicoid-serpiginous-geographic dystrophy.[9] All these can be distinguished on electrophysiological and morphological grounds. We hope that our cases will aid ophthalmologists and geneticists counselling other patients with this unusual condition.

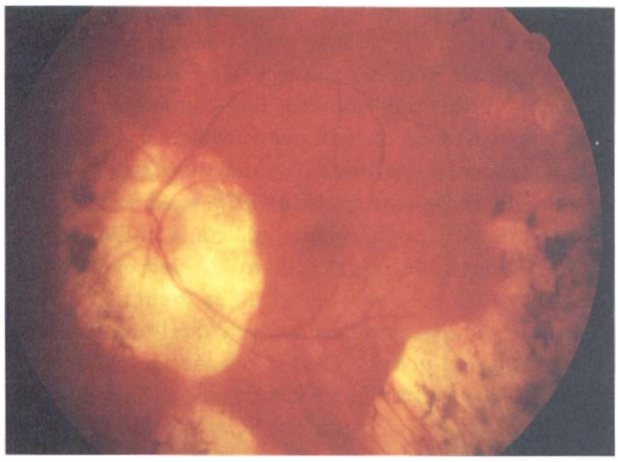

Fig. 3. *Case 2. Colour fundus photograph of the left eye showing peripapillary atrophy with atypical pigmentary changes.*

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