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Genetic isolate and preserved para-arteriole retinal pigment epithelium

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Abstract Preserved para-arteriole retinal pigment epithelium (PPRPE) is an uncommon form of retinitis pigmentosa, with a very peculiar fundoscopic appearance. To our knowledge no patient under age ten, affected by PPRPE, has been reported in the literature. The authors present here a seven-year-old boy, whose fundus examination is consistent with the diagnosis of PPRPE. The case report confirms that PPRPE starts early in childhood, and additionally supports the hypothesis of an autosomal recessive inheritance of this condition, since the proband's family lives in a 'genetic isolate'.

Key words Retinal pigment epithelium (RPE); retinitis pigmentosa (RP); preserved para-arteriole retinal pigment epithelium (PPRPE); genetic isolate; hyperopia.

Introduction There are specific types of retinitis pigmentosa (RP) with various morphological fundus patterns. Preserved para-arteriole retinal pigment epithelium (PPRPE) is an uncommon form of retinitis pigmentosa, first described by Heckenlively,¹ consisting of relative preservation of the retinal pigment epithelium (RPE) adjacent to and under the retinal arterioles.

The process starts in the anterior equator and peripheral regions of the retina, and with time the central areas of preserved RPE also succumb to the disease process. Preservation of macular RPE is usually observed in advanced RP; nevertheless, this specific pattern of RPE preservation adjacent to retinal arterioles has been reported very rarely in RP.

Unlike typical RP patients, who are myopic, these patients are often highly hyperopic. We present here the youngest patient affected by this condition, so far reported in the literature.

Case report The patient is a seven-year-old white boy, product of an uncomplicated pregnancy, with a reddish fair complexion. His past medical history was notable only for the usual childhood diseases. The family history was unremarkable for ocular problems. The parents were not consanguineous, but they lived in a 'genetic geographic isolate', and both were born in a small village in the Italian Alps.

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Fig. 1. Focal preservation of RPE adjacent to the arteriole with a diffuse loss of RPE.



The light complexion and the marked photophobia led the paediatrician to ask for an ophthalmological evaluation for suspected albinism when the child was 18 months old. At that time the examination revealed no significant problem apart from hyperopic defect.

Five years later, following a school screening visit, the child was sent to our University Department where examination disclosed a high hyperopic and astigmatic refractive error.

His cycloplegic refraction was RE 9.75-2.50 × 15 degree; LE 9.50-2.50 × 180 degree. At the time of our examination, the visual acuity was 20/70 in OU with correction [RE 9.25-2.50 × 15 degree; LE 8.75-2.25 × 180 degree]. Night blindness was noted by the parents, and, although not recognized by his parents, nystagmoid rotatory movements were present without any muscle imbalance at the time of our examination. Slit-lamp examination did not show transillumination of the iris, and the lenses were clear.

Fundus examination revealed a slightly pale optic disc with moderate attenuation of the retinal vasculature in both eyes, and a very hypopigmented fundus with general diffuse loss of RPE in the periphery (Fig. 1). Para-arteriole retinal pigment epithelium was preserved in anterior equatorial regions (Fig. 2).

Electrophysiological studies were abnormal. The ERG demonstrated a photopic waveform that was one third normal size, while the scotopic ERG was barely recordable, pointing to earlier or more severe involvement of the rod system in this condition. The reduction of the EOG light peak/dark ratio was expected because of the diminished ERG responses.

Blood analysis, urinalysis, phytanic acid levels, vitamin A and carotene levels, ornithine levels were all within the normal limits.

Discussion Preservation of posterior pole RPE is quite frequent in RP while preservation of para-arteriole RPE is uncommon.¹

Our subject had a pale optic disc with light attenuation of the retinal vasculature. The evidence of focal preservation of RPE adjacent to or under ar-

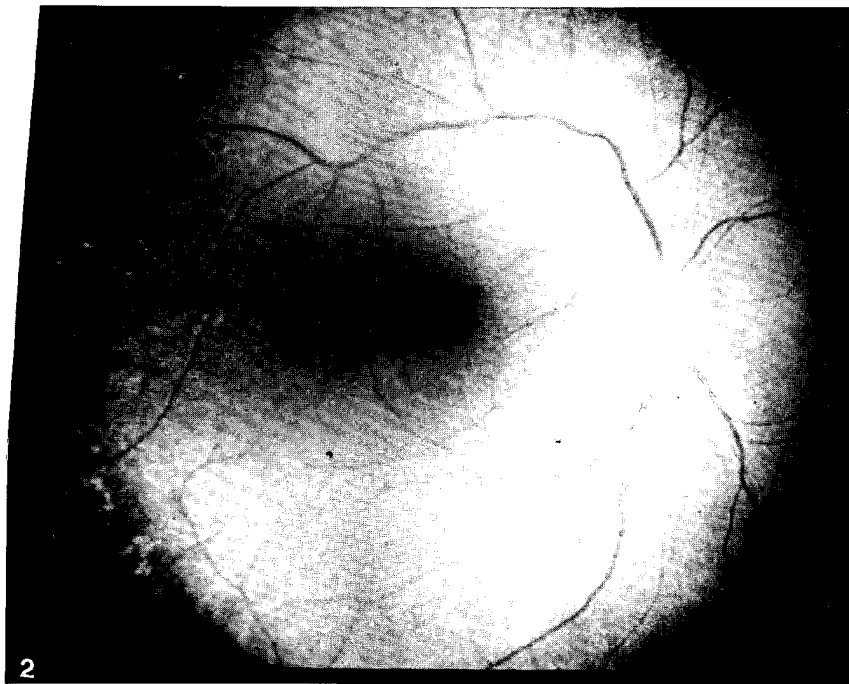


Fig. 2. Slightly pale optic disc with preservation of RPE in the anterior equator region.

terioles with a panretinal degenerative process makes our patient resemble those described in the literature by Heckenlively.¹

The focal preservation of the RPE in areas adjacent to arterioles implies a positive interaction between RPE and the arteriole. Many hypotheses have been put forward on the aetiology of the disease process, but they are not yet well known. Initially it was suggested that oxygenation from the arteriole may help to preserve the RPE, but this does not appear to be likely, since the RPE does not derive either its nutrition or oxygen from the retinal circulation.¹ Furthermore, any oxygenation effect is just as likely to come from the retinal capillary network as from the retinal arteriole.¹

Besides, it was assumed that this effect was due to a reduction to light exposure due to the shadow of the overlying vessels, but this theory is not supported by the same pattern under or adjacent to the veins. Examination of choroidal vasculature on fluorescein angiograms reported in the literature reveals no abnormalities of the choroidal circulation.^{1,2}

Some authors¹⁻³ have suggested that a permeable factor released by the retinal arteriole may have a positive influence in temporarily preserving RPE in the areas adjacent to these vessels. Gills and Wadsworth³ suggest that the primary aetiological process in these patients is in the bipolar layer, which receives nourishment from the retinal circulation, and that the retrograde degeneration could cause loss of the receptors and RPE, except in the proximity of arterioles. The reported features are consistent with the hypothesis that a permeable factor could be connected with PPRPE, locally retarding the degeneration in the areas of retinal arterioles.¹⁻³ It may be also possible that transsynaptic degeneration would explain the pale optic disc usually present in these patients.³ This issue remains controversial since the literature leans toward rebutting this possibility. To our knowledge, no patient younger than seven years of age with this type of RP has been reported in the literature.⁴

The absence of scattered pigment deposits or bone spicules in our seven-

year-old boy confirms the belief that these are typical late findings, and that the disease process starts in the periphery. Further evidence supporting the similarity of our case report to those reported in the literature, is the hyperopia, uncommon in patients with RP, who are usually myopic. This refractive error is a typical finding of another type of RP in which the disease is manifest at birth or very early in life: Leber's congenital amaurosis,^{5,6} but unlike Leber's form, in our patient nystagmus was not present before 18 months, when the child underwent the first ophthalmological visit.

In addition, the child's family lived in a small village in the Italian Alps, where both parents and grandparents were born. Geographic isolation could have altered the mutant allele frequency in that population, increasing the likelihood of occurrence of an autosomal recessive disease.

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