

© Ophthalmic Genetics 0167-6784/94/
US\$ 3.50

Ophthalmic Genetics – 1994, Vol. 15,
No. 3/4 pp. 161-164
© *Æolus Press*
Buren (The Netherlands) 1994

Accepted 28 September 1994

Macular dysplasia and pigmented paravenous retino-choroidal atrophy

Paolo Nucci
Maria Pia Manitto
Andrea Piantanida
Rosario Brancato

Department of Ophthalmology and Visual Sciences,
Scientific Institute S. Raffaele Hospital, University of Milan, Italy

Abstract Pigmented paravenous retino-choroidal atrophy (PPRCA) is a rare retinal disease characterized by bilateral patches of pigment and areas of chorioretinal atrophy distributed along the veins. The authors present a 21-year-old male with pigmented paravenous retinochoroidal atrophy and unilateral macular dysplasia. To their knowledge, this is the second reported case of macular involvement. They believe that such association is not occasional, but may be suggestive of a variable expressivity of the disease.

Key words Pigmented paravenous retino-choroidal atrophy; retinal pigment epithelium; macular dysplasia.

Introduction Pigmented paravenous retino-choroidal atrophy (PPRCA) is a rare retinal disease characterized by bilateral patches of pigment and areas of chorioretinal atrophy distributed along the veins. Most cases have been described in young male adults, and some authors^{1,2} have reported evidence of genetic transmission of this entity, although PPRCA occurrence is mostly sporadic. The etiology is unknown, however, inflammatory or dysgenetic causes have been proposed. This condition is usually asymptomatic and stationary, but a marked variability in the ocular findings of reported cases has been observed.¹⁻³ In the family reported by Traboulsi and Maumenee the severity of chorioretinal changes ranged from a few coarse paravenous pigmented clumps to diffuse chorioretinal atrophy.² The wide variability of the clinical features suggests a heterogeneous spectrum of conditions.

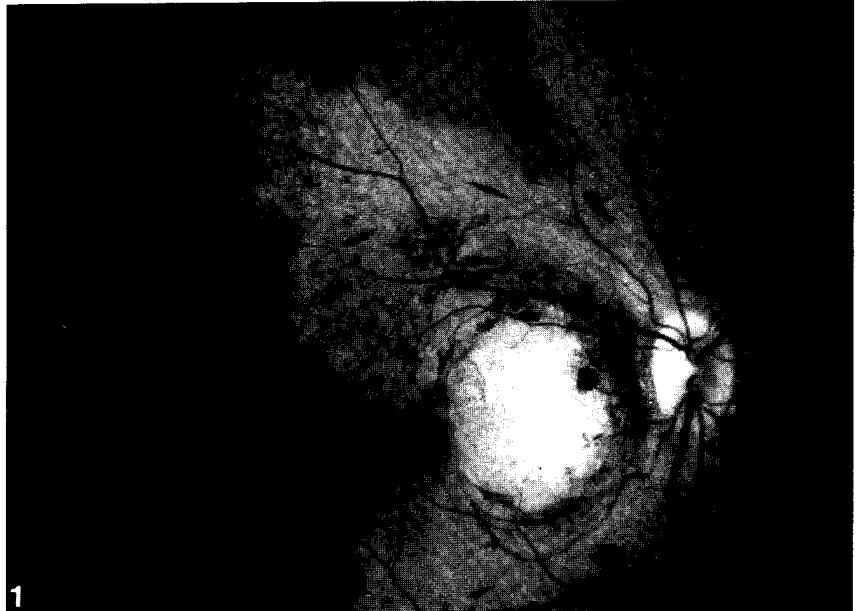
We examined a 21-year-old male with pigmented paravenous retinochoroidal atrophy and unilateral macular dysplasia. Based on the knowledge presently available in the literature, this is the second reported case of macular involvement. A similar ocular finding has been previously described by Traboulsi and Maumenee in one patient of their family with PPRCA.²

We believe that such an association is not occasional, but may be suggestive of a variable expressivity of the disease.

Correspondence to: Paolo Nucci, M.D.,
Department of Ophthalmology and
Visual Sciences, Scientific Institute S.
Raffaele Hospital, University of Milan,
Via Olgettina 60, 20132 Milan, Italy.
Fax +39-2-2643 2482

Acknowledgements: The authors are
grateful to Professor Paliaga for
referring the patient.

Fig. 1. Fundus photograph: right eye, macular 'colobomata' and slight scleral ectasia.



Case report A 21-year-old Caucasian male was referred to our department for evaluation of retinal degeneration. His family history was not significant with regard to ocular problems, and his past ophthalmological history consisted of a visit for esotropia when the patient was six years old. At that time the visual acuity was counting fingers in RE and 20/25 in LE with a slight hyperopic astigmatism (+1 -1 × 180). The deviation measured 30^Δ of esotropia and 7^Δ of right hypertropia. Atrophic macular changes were described in the right fundus with a helicoidal chorioretinal atrophy associated with bone spicule pigmentation in both eyes. The ophthalmological examination was otherwise normal.

Fig. 2. Fluorescein angiography: left eye, transit phase: paravenous chorioretinal atrophy and coarse pigmentary changes along the veins.



Our examination showed similar functional data. We found normal pupillary function and absence of afferent pupillary defect in both eyes. Visual field and color vision (Ishihara) were normal in the LE, and were considered unreliable in the RE.

Fundus examination disclosed, in the RE, no abnormality of the vitreous body, a normal optic disc and a macular coloboma, with a slight scleral ectasia (Fig. 1); paravenous chorioretinal atrophy with coarse pigmentary changes and attenuated retinal arterioles were evident in both eyes (Fig. 2). Fluorescein angiography showed extensive transmission defects along the veins. The described findings were consistent with the diagnosis of PPRCA.

Electroretinography showed decreased photopic and scotopic responses and the electro-oculography was subnormal: light-peak-to-dark-trough ratio was 1.47 for the RE and 1.43 for the LE.

The patient was not the product of a consanguineous marriage, and none of the other family members had any fundus change. The results of systemic and laboratory examinations (blood pressure, hematologic evaluation, blood chemistry studies, urinalysis, chest roentgenogram) were all in the normal range. Toxoplasma, syphilis and CMV titers, as well as a skin test for tuberculosis were also negative.

Discussion PPRCA, first described by Brown in 1937, presents peculiar ocular findings consisting of paravenous choroidal atrophy and bilateral patches of pigment along the veins.¹ Patients are usually asymptomatic and the diagnosis is based on the typical fundus picture. However, the clinical features associated with this condition show considerable heterogeneity among reported cases. The condition commonly affects young adults, but Hayasaka³ described a patient diagnosed at 68 years of age. Only two cases of familial occurrence, compatible with dominant or X-linked transmission, have been reported.^{1,2}

In the family reported by Traboulsi and Maumenee the characteristic fundus abnormalities were associated with hyperopia, esotropia, vitreoretinal degeneration and subtle systemic findings in the form of bifid uvulae or mild loose-jointedness.²

The retinal findings in most patients are stationary and the condition, often asymmetric, is nonprogressive or slowly progressive in nature. All these aspects point to a 'local' developmental defect. Despite this, Pearlman *et al.* were the first to show ophthalmoscopic and functional progression.⁴ Visual loss was reported in one patient.²

A wide variability exists also among electrophysiological tests. Skalka's¹ and Hagasaka's³ electroretinographic studies revealed subnormal responses under both photopic and scotopic conditions, consistent with the findings in our patient. Traboulsi and Maumenee noted normal b₂-wave studies and decreased b₁-wave amplitudes.² Electro-oculography showed variable Arden test values ranging from normal to borderline¹ and abnormal, as in the herein reported case.

On fluorescein angiography, extensive transmission defects are visible, indicating loss of the retinal pigment epithelium as well as choriocapillaris along the retinal veins. Skalka¹ noted differences according to the age of the patients, in the form of preservation of chorioidal vasculature underlying the atrophic retinal pigment epithelium in younger subjects. Such relative sparing of the choriocapillaris in young patients suggests that the disease involves the

retinal pigment epithelium first, with secondary atrophy of underlying chorioidal vasculature.

It is likely that this condition has multiple etiologies. Some authors stress the role of genetic factors in its pathogenesis, although an inflammatory or infectious cause has also been proposed. Skalka believed that PPRCA may be considered another incomplete, self-limited form of retinitis pigmentosa. The angiograms of our patient disclosed an alteration of the blood-retinal barrier, at the level of the retinal pigment epithelium, similar to that shown in persons with retinitis pigmentosa.⁵

The most interesting feature of our patient is the unilateral macular dysplasia. Traboulsi and Maumenee² reported a patient with an excavated macula, a finding consistent with Mann's classification of macular 'colobomata'.⁶ It may not be possible to distinguish between inflammatory and developmental etiology of this lesion, on clinical appearance alone, but we believe that a failure in the differentiation that takes place from the eighth month of intra-uterine life to the fourth month after birth, could possibly explain the macular anomaly.

References

- 1 Skalka HW. Hereditary pigmented paravenous retinochoroidal atrophy. *Am J Ophthalmol* 1979, 87: 286-291.
- 2 Traboulsi EI, Maumenee IH. Hereditary pigmented paravenous chorioretinal atrophy. *Arch Ophthalmol* 1986, 104: 1636-1640.
- 3 Hayasaka S, Shibasaki H, Noda S, Fujii M, Setogawa T. Pigmented paravenous retinochoroidal atrophy in a 68-year-old man. *Ann Ophthalmol* 1991, 23: 177-180.
- 4 Pearlman JT, Heckenlively JR, Bastek JV. Progressive nature of pigmented paravenous retinochoroidal atrophy. *Am J Ophthalmol* 1978, 85: 215-219.
- 5 Newsome DA. Retinal fluorescein leakage in retinitis pigmentosa. *Am J Ophthalmol* 1986, 101: 354-360.
- 6 Mann IC. On certain abnormal conditions of the macular region usually classified as colobomata. *Br J Ophthalmol* 1927, 11: 99-116.