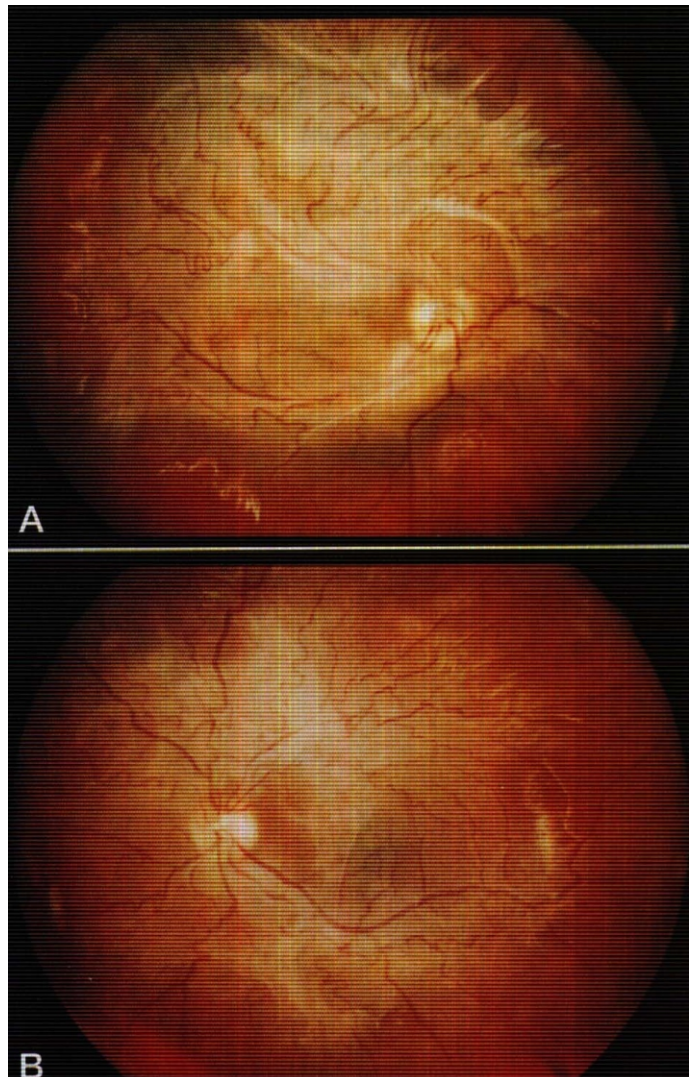


# COMBINED HAMARTOMA OF THE RETINA AND RETINAL PIGMENT EPITHELIUM: A Bilateral Presentation

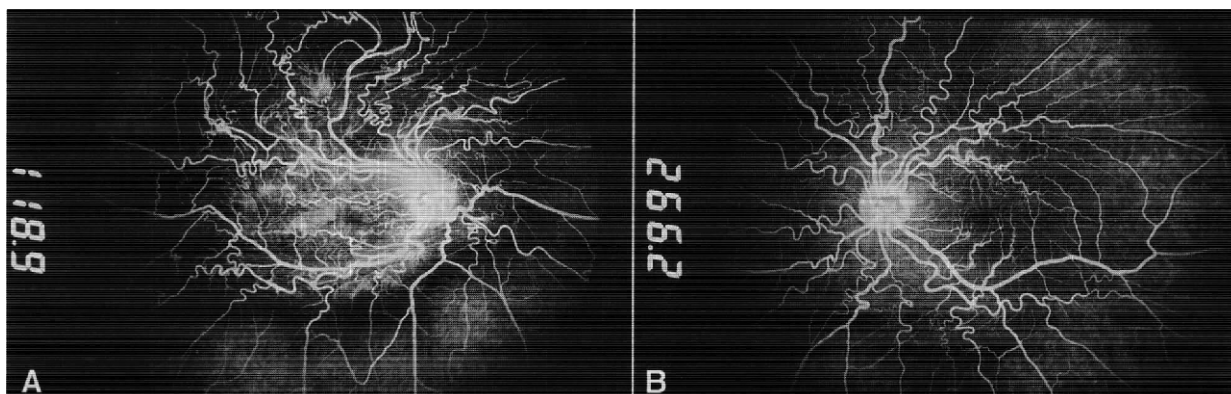
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Combined hamartoma of the retina and the retinal pigment epithelium (CHR-RPE) is a rare developmental disorder assumed to be present at birth.<sup>1</sup> The significance of this rare entity lies primarily in its resemblance to malignant conditions and in the visual



**Fig. 1.** Color picture of the fundus of the right (A) and left (B) eyes. Note that almost all of the fibrous tissue is subretinal or intraretinal, at the level of the retinal pigment epithelium and within the neural retina. The epiretinal component is manifested mainly in tortuosity of the retinal vessels.



**Fig. 2.** Fluorescein angiography of the left eye (**B**) manifests prominent tortuosity of blood vessels, traction of the preretinal membrane on some retinal vessels, and moderate leakage from the disc. The right eye (**A**) manifests, in addition, multiple spots of subretinal leakage.

reduction it induces. Diagnosis is based on characteristic clinical findings, aided by fluorescein angiography and echography.

In the majority of cases the condition remains stable, both functionally and morphologically, throughout life. Patients should, however, be followed up; the epiretinal component may progress, cause further reduction in vision, or lead to a tractional-type retinal detachment.

Almost all published cases were unilateral. Of the

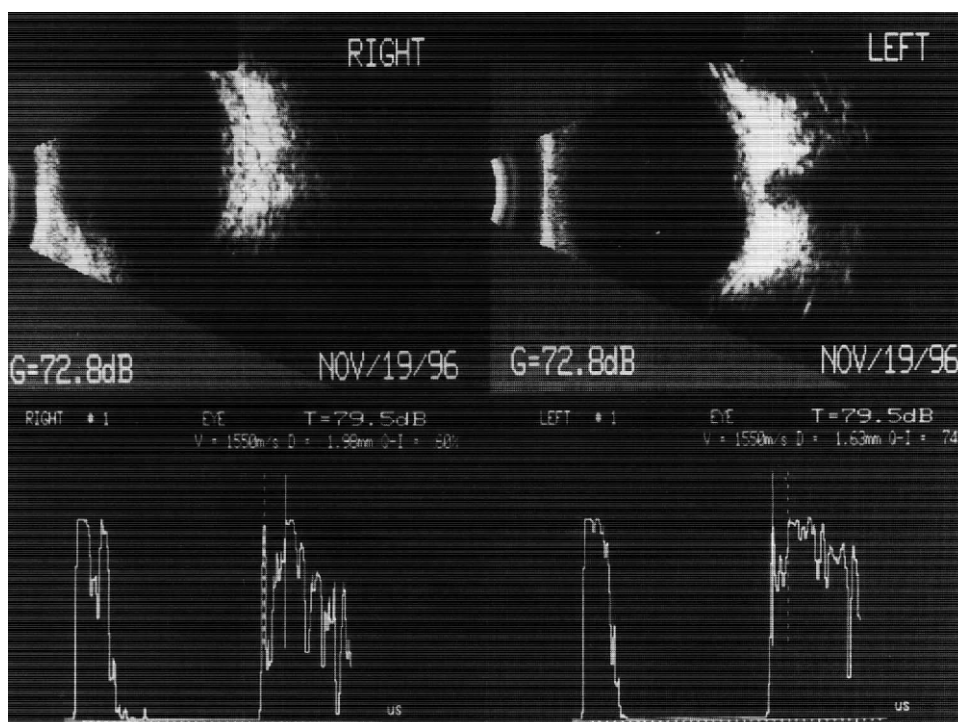
60 cases compiled by members of the Macula Society,<sup>1</sup> none manifested bilaterally. A few bilateral cases have been previously reported.<sup>2,3</sup> An association between neurofibromatosis and bilateral retinal hamartomas has been described rarely.<sup>4,5</sup>

We report an 8-year-old boy with a bilateral, unusual, and fairly symmetrical involvement of CHR-RPE.

### Case Report

An 8-year-old boy was referred to us with a history of poor vision since the age of 2 years. Medical and family history were otherwise unremarkable. No clinical stigmata of neurofibroma-

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**Fig. 3.** Standardized echography of the posterior pole lesions. **Top**, vertical macular B-mode scans. **Bottom**, standardized A-mode at tissue sensitivity. **Left**, right eye. **Right**, left eye. Note that at least two layers are affected.

## BRIEF REPORTS

tosis was present. Corrected visual acuity was 6/60 in the right eye and 6/24 in the left. In each fundus a mildly elevated, pigmented, and somewhat greenish-gray macular lesion was present. In both eyes the lesion spanned the entire macular region, beyond the temporal arcades, as well as encompassing the disc and peripapillary region (Figure 1). Ophthalmoscopically, preretinal fibrosis exerting traction on the retina was seen, evident as retinal folds and prominent vessel tortuosity and dragging. In addition, a much more extensive subretinal fibrosis at the level of the pigment epithelium was noted. Fluorescein angiography demonstrated pronounced tortuosity of retinal vessels in both eyes and leakage spots within the lesion in the right eye (Figure 2).

B-scan echography revealed elevated lesions at the macular area in both eyes with an irregular inner surface. Standardized A-mode characterized the lesions as elevated, solid, low to medium, and irregularly reflective lesions, with a maximal measured elevation of 2 mm in the right eye (Figure 3).

### Discussion

This case substantiates the existence of a bilateral variant of CHR-RPE. The history of reduced vision since early childhood is consistent with the assumption that these lesions are congenital. Also of interest is the fact that bilateral cases of CHR-RPE usually present in a rather symmetrical distribution of the lesions. It is difficult to establish the correct diagnosis in such a case. The diagnosis in our patient was mainly based on the clinical finding of several layers (RPE, neural retina, and epiretina) being separately affected by a pathologic layer of gliosis. It was further substantiated by the early onset of the symptoms and the separate vascular layers seen by fluorescein angiography and ultrasound.

### References

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