Acquired Combined Hamartoma of the Retina and Retinal Pigment Epithelium

The etiology of combined hamartoma of the retina and retinal pigment epithelium remains speculative, although the consensus has been that it is a congenital lesion present at birth, as inferred from the relatively early presentations of reported cases. We describe a patient with a combined hamartoma that developed from a focus of apparently normal retina, with concurrent serial fundus photographs.

Report of a Case | A 6-month-old girl with an unremarkable medical and family history was referred for examination under anesthesia for a total tractional and exudative retinal detachment of the right eye. Findings on clinical examination of the left eye were unremarkable (Figure 1A). Widefield fluorescein angiography showed disorganized vascular branching in the right eye, with full vascularization in the left eye and normal posterior pole and midperipheral vasculature (Figure 1B). There were a total of 2 clock hours of atypical vascular patterns in the left eye in the far periphery, but with full perfusion. A diagnosis of familial exudative vitreoretinopathy was made, and the patient underwent surgery in the right eye.

Examination under anesthesia 2 months postoperatively showed improvement in the right eye and continued normal findings on examination in the left eye. However, 3 months later, repeated examination of the left eye under anesthesia revealed a discrete lesion that was round, mildly raised, dark charcoal gray, and nasal to but not involving the disc, which was not present on prior examinations (Figure 2A). Periretinal glial proliferation was present over the surface, and the surrounding retinal vasculature was contracted toward the lesion. Fluorescein angiography confirmed a tumor at the level of the retina and retinal pigment epithelium (Figure 2B), consistent with a presumed combined hamartoma of the retina and retinal pigment epithelium. Optical coherence tomography and ultrasonography were not performed, although they are not required for the diagnosis. Continued observation for 2 months more showed subtle growth of the tumor with additional gliosis.

Discussion | Gass described 7 patients with combined hamartoma and synthesized his findings with previous reports, establishing a new clinical entity. He proposed that “they probably are present at birth, and they probably represent focal areas of maldevelopment of the pigment epithelium, retina, blood vessels, and overlying vitreous.” The youngest patient in his series was 19 months old. In the Macula Society’s subsequent collaborative report, the mean age was 15 years and the youngest patient was aged 10 months. Shields et al described the youngest patient at age 2 weeks. This trend of early presentations seemed to validate Gass’ inference that these are congenital lesions present at birth.

In contrast, we report a case in which a presumed combined hamartoma developed after birth—seemingly de novo—between ages 8 and 11 months. Although there are a few reports of acquired combined hamartomas, these previous cases involved distinct insults to regions that involved or were contiguous with areas that eventually developed hamartomas, such as optic disc edema and vitreomacular traction—suggesting, rather, that these lesions were acquired hamartoma-like reactive gliosis and hyperpigmentation. Our patient has
mild familial exudative vitreoretinopathy in the left eye, but the mild vascular changes are in the far periphery, well separated from the hamartoma, and the area that developed the lesion was fully vascularized and free of macroscopic damage or other inciting primary pathology.

We cannot, of course, completely discount the influence of familial exudative vitreoretinopathy as presumed global wnt signaling aberrations, which, although unlikely, may have altered the timing of the hamartomatous growth. Additionally, we do not infer that all combined hamartomas are acquired; in considering the series by Shields et al, it is more likely that a hamartoma in a 2-week-old patient was indeed present at birth.

In summary, we report a presumed combined hamartoma that was acquired after birth in a child whose fellow eye's presentation allowed incidental observation of its development.

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Retrospective Appraisal of Split-Cornea Transplantation: An Audit of 1141 Donor Corneas

For more than 100 years, penetrating keratoplasty (PK) with full-thickness replacement of a diseased cornea with an allograft donor tissue has been successfully performed. Especially within the last 10 years, surgical techniques have been improved to the point that selective replacement of the diseased structure of the cornea is possible. Diseases of the corneal endothelium (eg, Fuchs endothelial dystrophy, pseudophakic bullous keratopathy) are the main reasons for corneal transplantations and can be managed by Descemet stripping automated endothelial keratoplasty or Descemet membrane endothelial keratoplasty (DMEK),1,3,4 Deep anterior lamellar keratoplasty (DALK) allows selective replacement of the anterior pathologic corneal tissue (eg, advanced keratoconus, herpetic corneal...