A Case of a Combined Hamartoma of the Retina and RPE Concurrently Found in the Same Eye as Peripheral Astrocytic Hamartomas

Stefanie D. Sherman, MD, Sidney A. Schechet, MD, Michael J. Shapiro, MD, Michael P. Blair, MD

Abstract
We report a case of a 7-year-old boy with neurofibromatosis 2 who was referred to our pediatric retina clinic for deteriorating vision of his left eye. Fundoscopy revealed a gray epiretinal and vitreous opacity extending to the fovea in the left eye. Optical coherence tomography (OCT) demonstrated an epiretinal membrane (ERM) with a distinct tissue plane and overlying vitreoretinal strands, consistent with combined hamartoma of the retina and retinal pigment epithelium (CHRRPE). Intraoperative OCT-guided pars plana vitrectomy and membrane peel were performed and the tissue was sent for histopathologic evaluation. Intraoperative scleral-depressed examination also revealed multiple elevated, white-gray, semitranslucent masses near the ora serrata, consistent with astrocytic hamartomas. Histopathologic analysis of the epiretinal tissue revealed glial origin. Recognition of any of these lesions in a young patient should prompt neurologic and genetic evaluation.