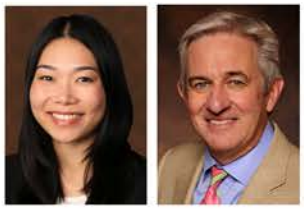
A 42-Year-Old Male with a Tumor at the Optic Disc and Macula

Elysse Tom, MD; Thomas K. Krummenacher, MD



Introduction:

A 42-year- old male presented to our clinic with persistent blurry vision, occasional persistent floaters, and an occasional pressure sensation in the right eye. He was referred to our clinic by optometry for an optic nerve and retinal lesion. He was initially seen at our clinic at the age of 24 and was then lost to follow up. Visual acuity was 6/200E in the right eye and 20/20 in the left eye. Intraocular pressures were normal in both eyes. There was no relative afferent pupillary defect. Anterior segment exam was unremarkable. Fundus exam revealed a hypopigmented and hyperpigmented peripapillary elevated lesion with an epiretinal membrane in the right eye.

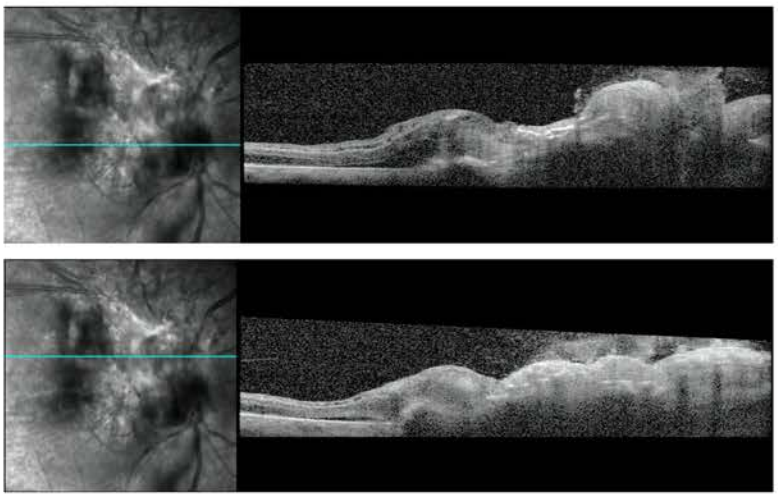


Figure 1: Optical coherence tomography of the macula shows opacities in the vitreous, an ERM, thick, disorganized retinal layers and peripapillary subretinal fluid.

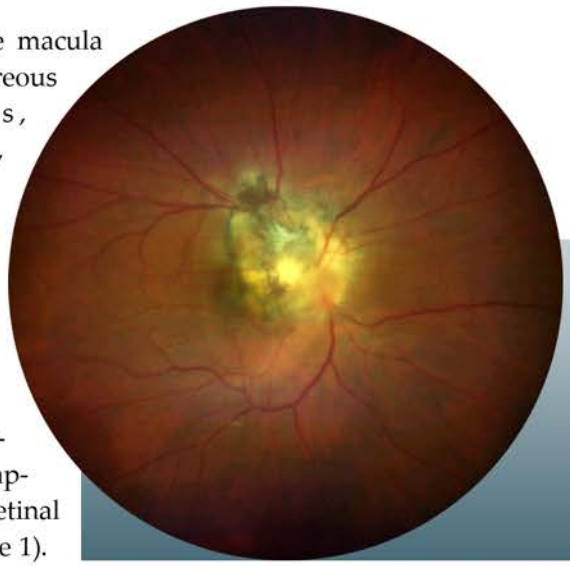
To aid in the diagnosis, an OCT and widefield fundus photos were obtained.

OCT of the macula showed vitreous opacities, thickened, disorganized retinal layers, an epiretinal membrane, and a small sliver of peripapillary subretinal fluid (Figure 1).

Photos showed an elevated lesion involving the optic nerve and superonasal macula with varying pigmentation and an epiretinal membrane (Figure 2). The tumor appeared consistent with a combined hamartoma of the retina and retinal pigment epithelium (CHRRPE).

Discussion:

Combined hamartoma of the retina and RPE is a benign tumor composed of pigmented epithelial cells, vascular tissue, and glial cells that may cause significant vision loss depending on its location. It is usually found at the optic disc or macula and is usually unilateral and solitary. CHRRPE is most often seen in children and may progress over time [1].



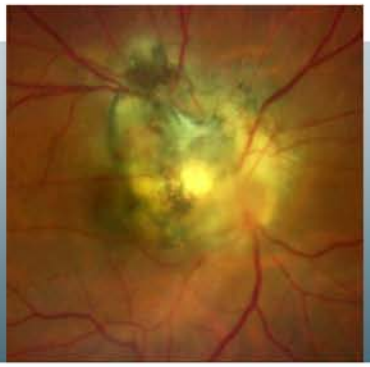
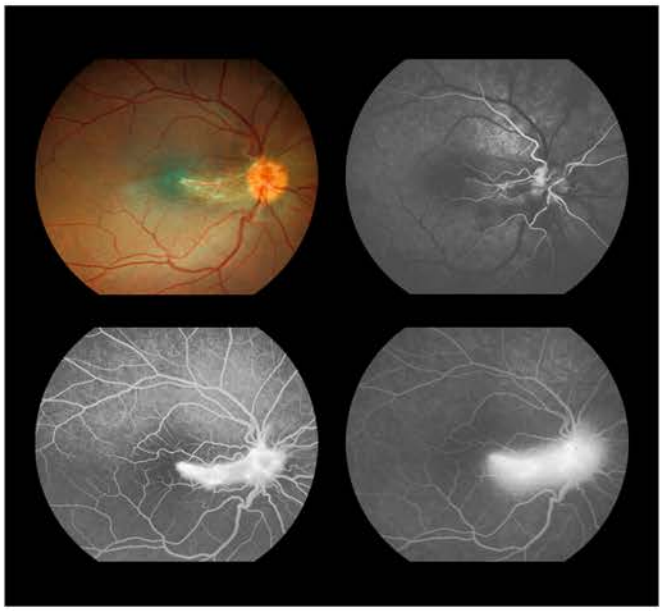


Figure 2a: Widefield photo shows the multi-pigmented lesion involving the optic disc and superonasal macula.

Figure 2b: Detail of the lesion.



Images (both left and above): Yannuzzi, et al. The Retinal Atlas. Second edition. 2017.

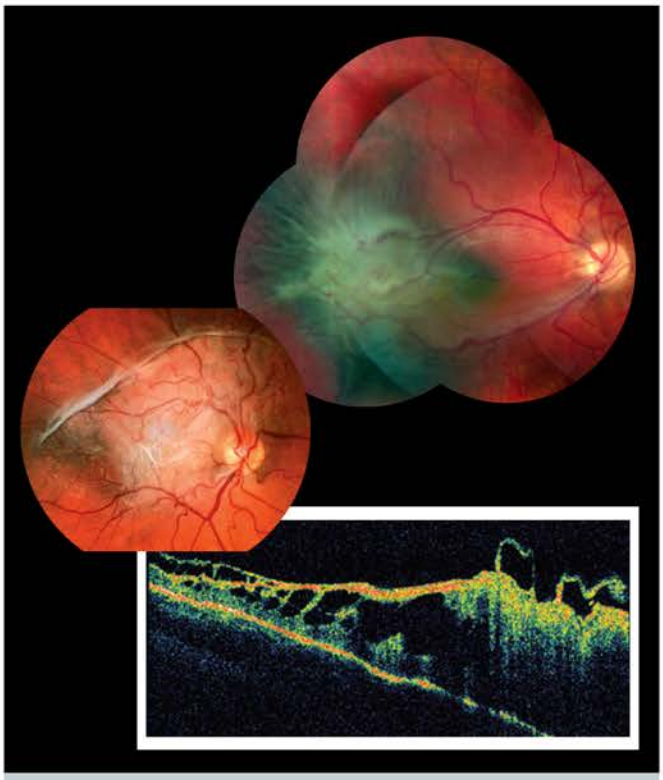
The etiology is unknown, but it is thought to be a congenital lesion that arises from undifferentiated cells initially intended to be RPE cells [2]. The term CHRRPE was first used by Gass in 1973 [3].

Patients usually present with decreased vision (40%) and strabismus (28%), but may also present with floaters, ocular pain, and leukocoria. Vision is worse than 20/200 in about 50% of cases at presentation [1]. Age of presentation ranges from 1 year of age to young adulthood. Exam reveals an elevated lesion with varying pigmentation, vascular tortuosity, and epiretinal membrane formation. Fundus exam may also show exudation, gliosis, corkscrew vessels, macular edema, vitreous hemorrhage, retinal detachment, neovascularization, macular hole, or peripheral hole formation [1, 4]. Most patients do not have systemic disease but there are reports of an association with neurofibromatosis 1 and 2, especially in bilateral cases [5, 6].

On ancillary imaging, optical coherence tomography usually reveals an epiretinal membrane with disorganized retinal layers and adjacent normal retina [7]. Fluorescein angiogram exhibits early hypofluorescence due to blockage by pigmented epithelial cells and late leakage from dilated vessels [4].

Differential diagnosis for tumors of the RPE, pigmented posterior segment tumors, and tumors near the optic disc also includes congenital simple hamartoma of the RPE, melanocytoma, choroidal melanoma, astrocytoma, RPE adenoma or adenocarcinoma [8]. In pediatric patients it is also important to consider retinoblastoma, persistent hyperplastic primary vitreous, toxocariasis, and morning glory disc anomaly. CHRRPE should also be differentiated from congenital hypertrophy of the RPE (CHRPE) which is also a hamartoma but is flat, sharply demarcated, and has a normal appearance of the overlying retinal and vessels [9].

In managing CHRRPE, it is important to attempt to prevent amblyopia if patient presents at a young age [4]. An ERM peel may be attempted but reports regarding improvement in visual acuity are a topic of debate [10, 11, 12]. Laser treatment or anti-vascular endothelial growth factor injections may be attempted if there is a choroidal neovascular membrane [13]. The patient should be followed every 6 months to monitor for progression of the lesions or complications such as epiretinal membrane, macular edema, choroidal neovascular membrane, retinal detachment, or retinal holes.



Images: Yannuzzi, et al. The Retinal Atlas. Second edition. 2017.

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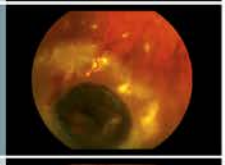
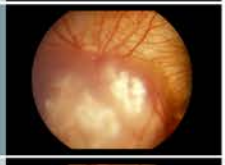
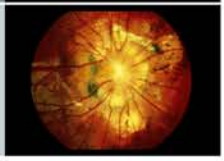
14. Yannuzzi, Lawrence A., et al. The Retinal Atlas. Second edition. [Philadelphia]: Elsevier, 2017.

Differential Diagnosis Table [14]

Congenital simple hamartoma of the RPE	
Melanocytoma	
Melanoma	
RPE adenoma or adenocarcinoma	
Astrocytic hamartoma	
Retinoblastoma	
Toxocariasis	
Persistent hyperplastic primary vitreous	
Morning glory disc anomaly	

All images in the differential diagnosis table are from The Retinal Atlas by Yannuzzi et al (14).

Case of the Month Supported by:

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