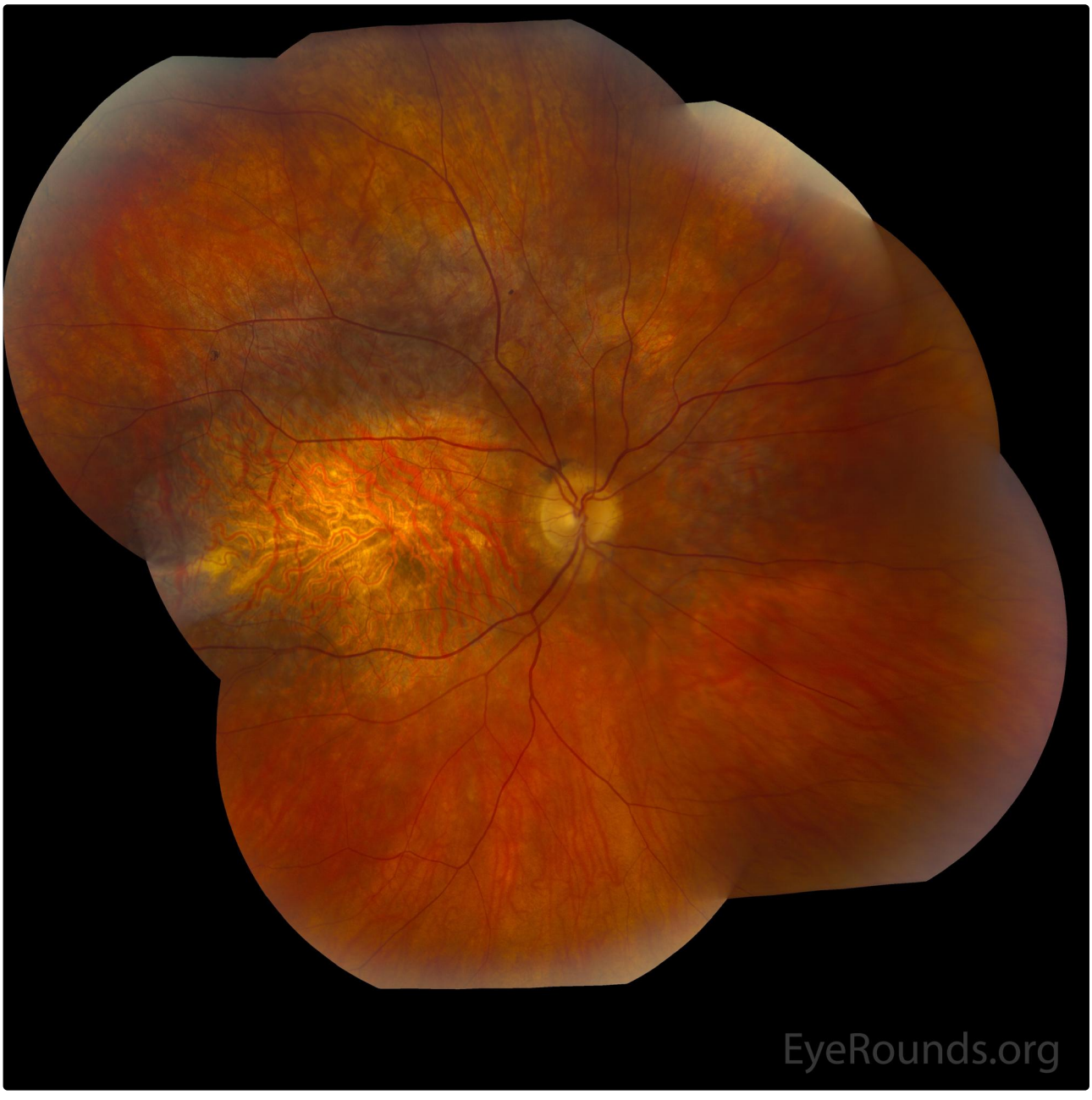
Pattern dystrophy

Category(ies): Inherited Eye Diseases, Retina/Vitreous

Contributor: Aaron M. Ricca, MD

Montage color fundus photographs are from a 63-year-old patient with molecularly-confirmed pattern dystrophy, which is an autosomal dominant, inherited retinal degeneration due to a mutation in the RDS/peripherin gene located on chromosome 6. Pattern dystrophy presents with a varying appearance of lipofuscin deposition and retinal atrophy with retinal pigment epithelial changes in the central macula, as demonstrated above. The disease demonstrates variable expressivity, and macular findings range from subtle to striking. Patients should be monitored with dilated fundus examination, as they are at risk for developing a choroidal neovascular membrane.



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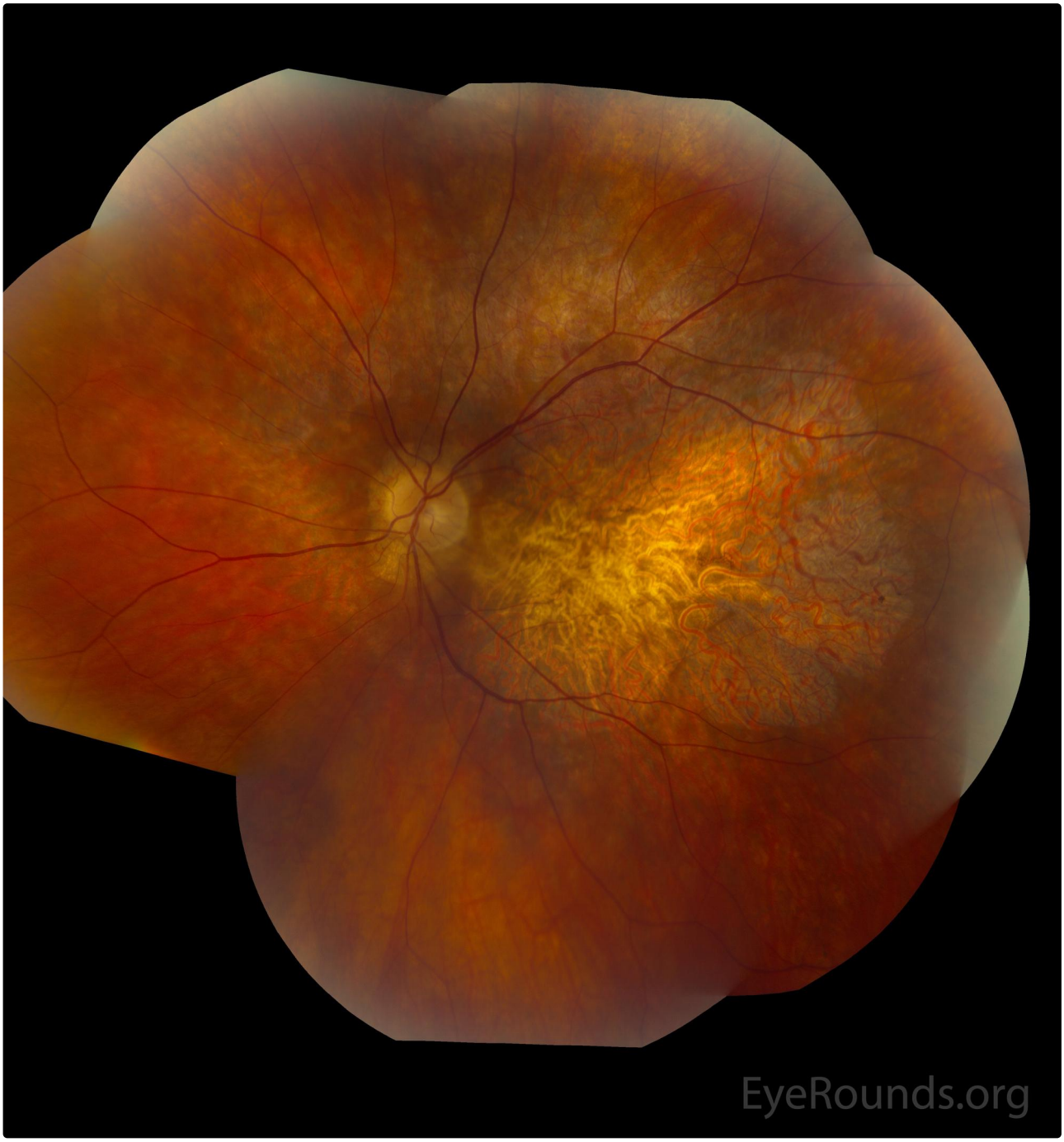

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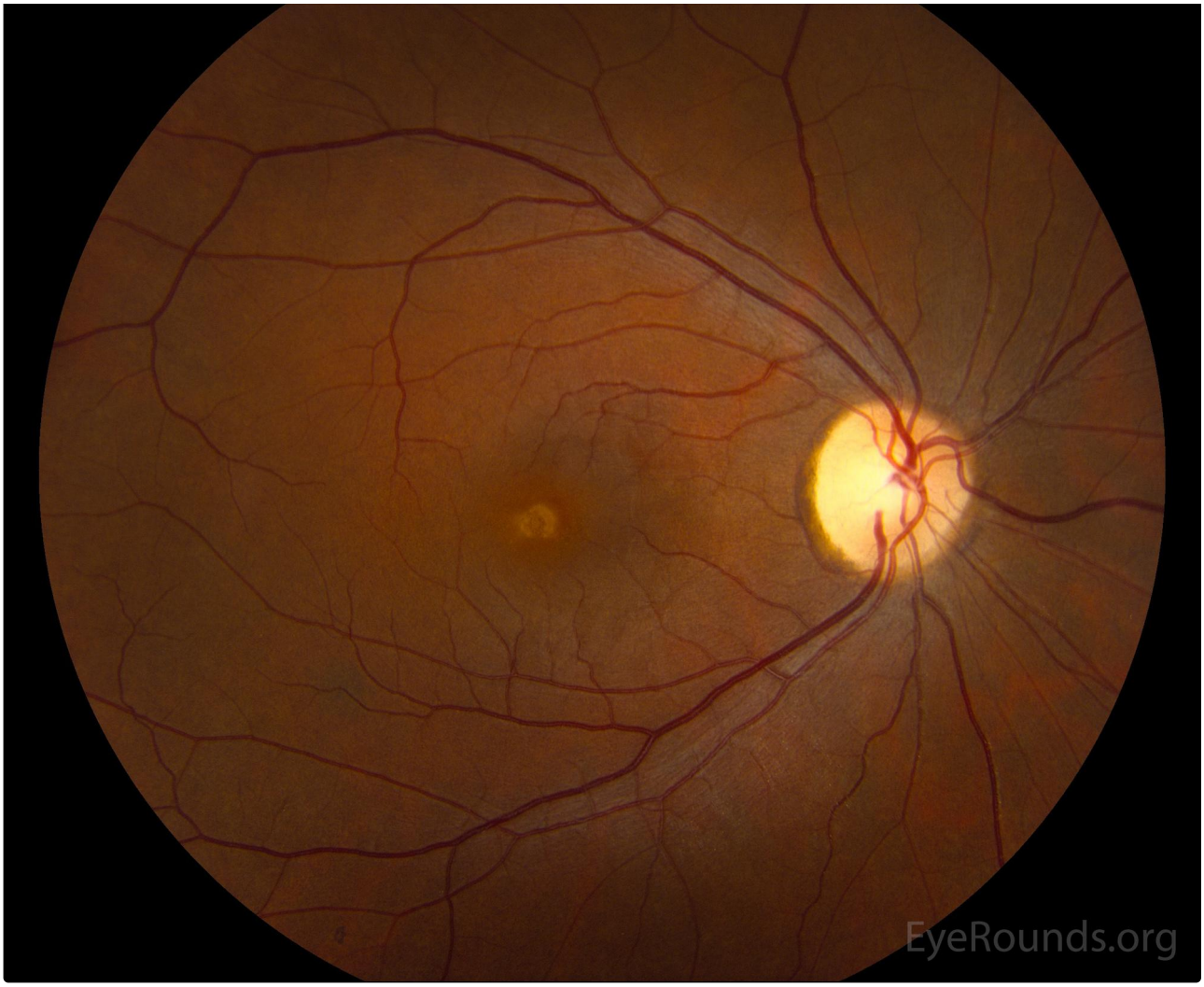
Contributor: Aaron M. Ricca, MD

Images are from a 42-year-old female with molecularly-proven pattern dystrophy. A mutation in the RDS/peripherin gene causes pigmentary changes at the level of the retinal pigment epithelium in various fundus "patterns." These different phenotypes are often grouped into the following types of dystrophies: butterfly-shaped, adult-onset vitelliform, peculiar foveomacular, reticular, and fundus pulverulentus. The above patient is an example of adult-onset vitelliform dystrophy. The fundus photos show the small foveal vitelliform lesion, and the optical coherence tomograms demonstrate the subretinal hyper-reflective material, without concern for a neovascular membrane.



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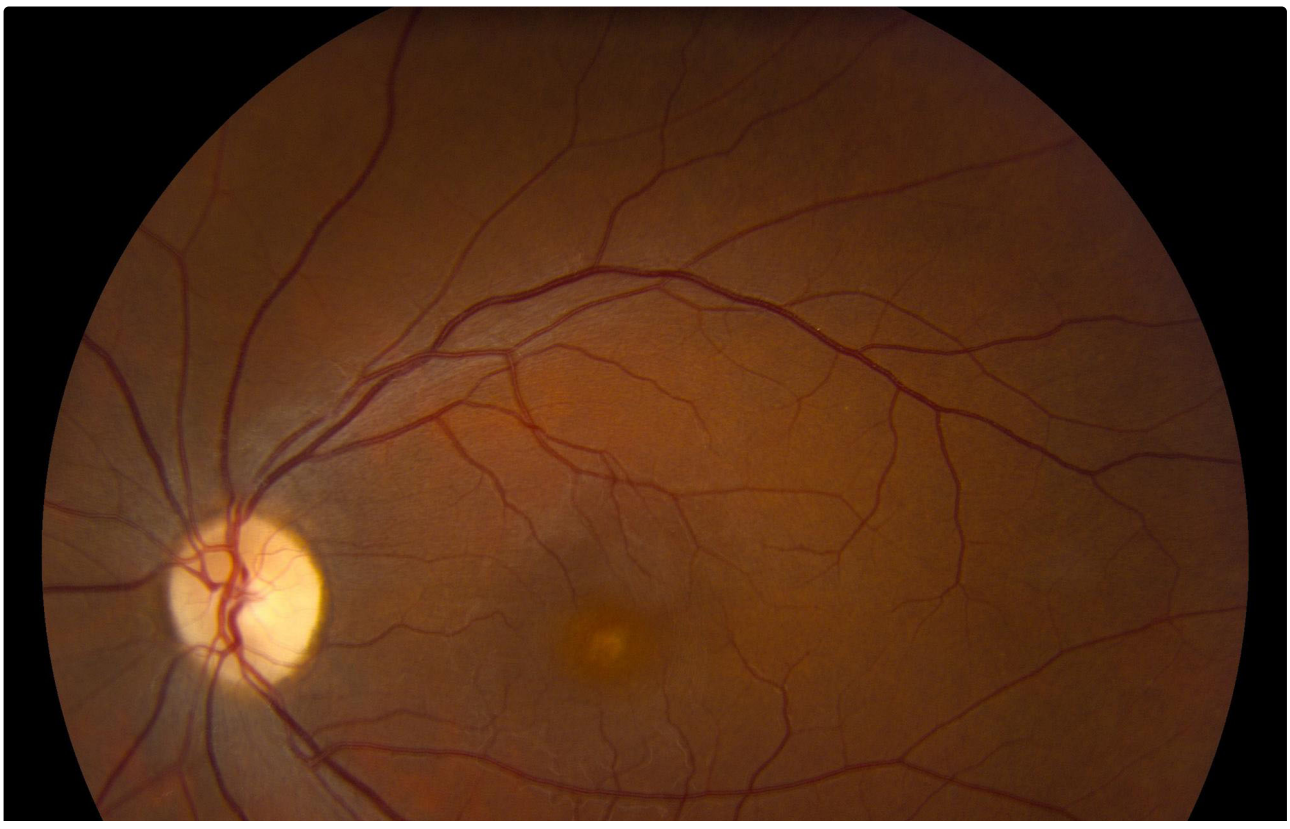

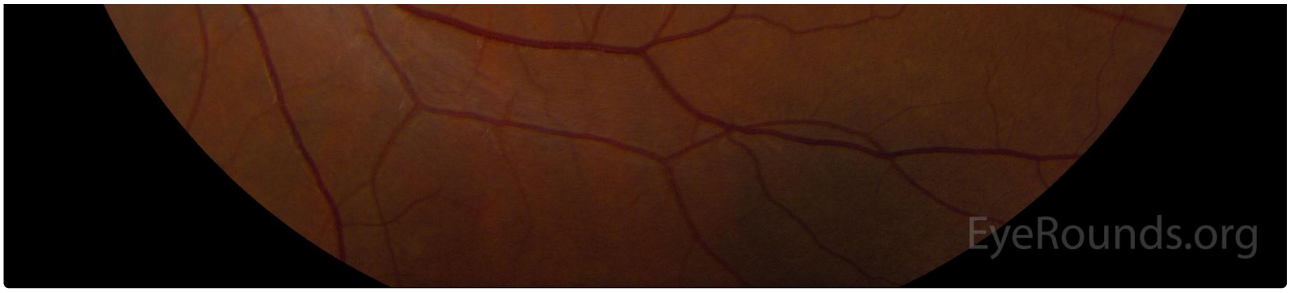

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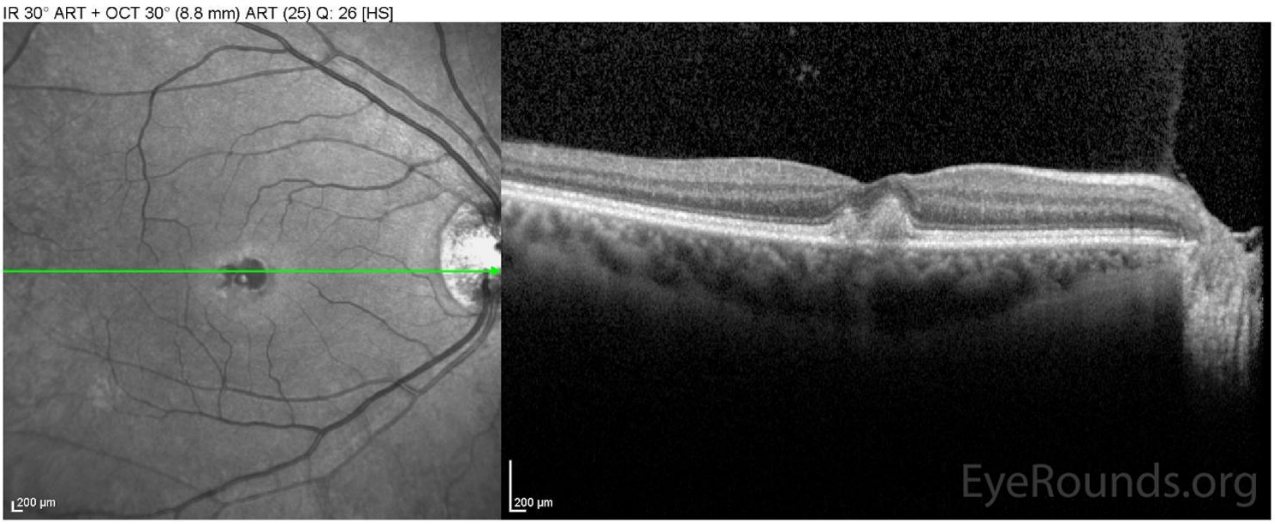

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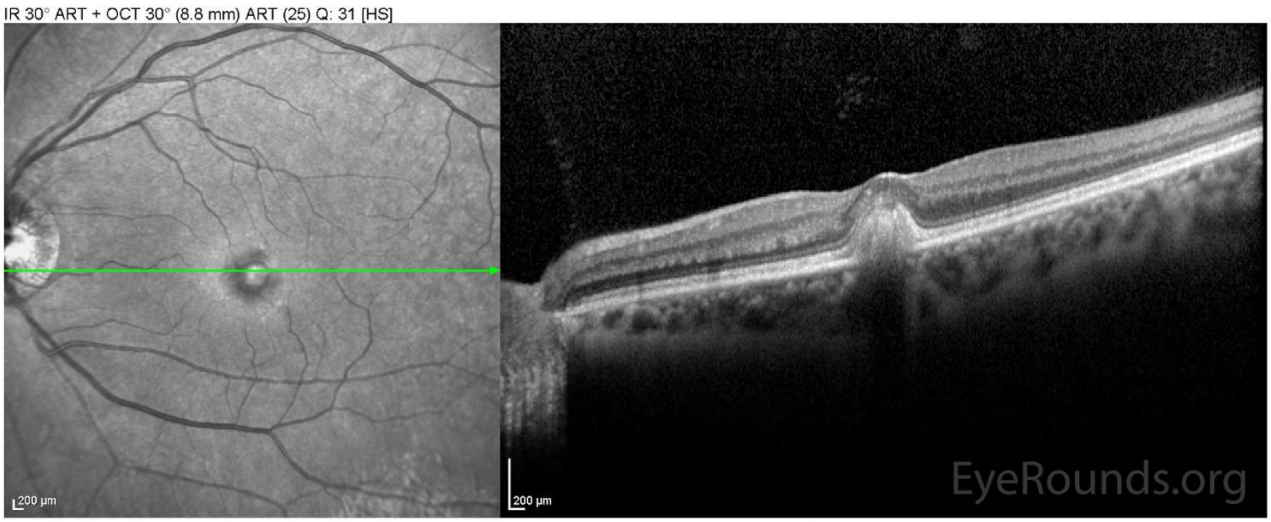

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Contributor: Andrew Doan, MD, PhD

This is a 40-year-old patient with a diagnosis of pattern dystrophy. The patient has good vision (20/25 in both eyes) and symmetric foveal retinal pigmented epithelium (RPE) hyperpigmentation in a butterfly pattern in both eyes.

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Reference:

Ryan SJ. Retina. 5th ed. London: Saunders/Elsevier; 2013.

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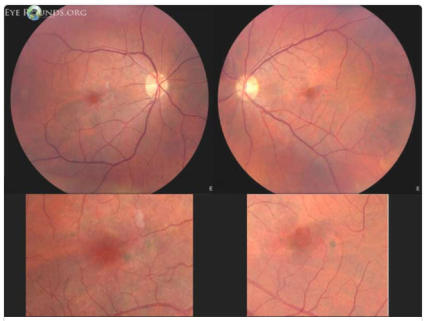

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