Retinitis pigmentosa (RP)

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

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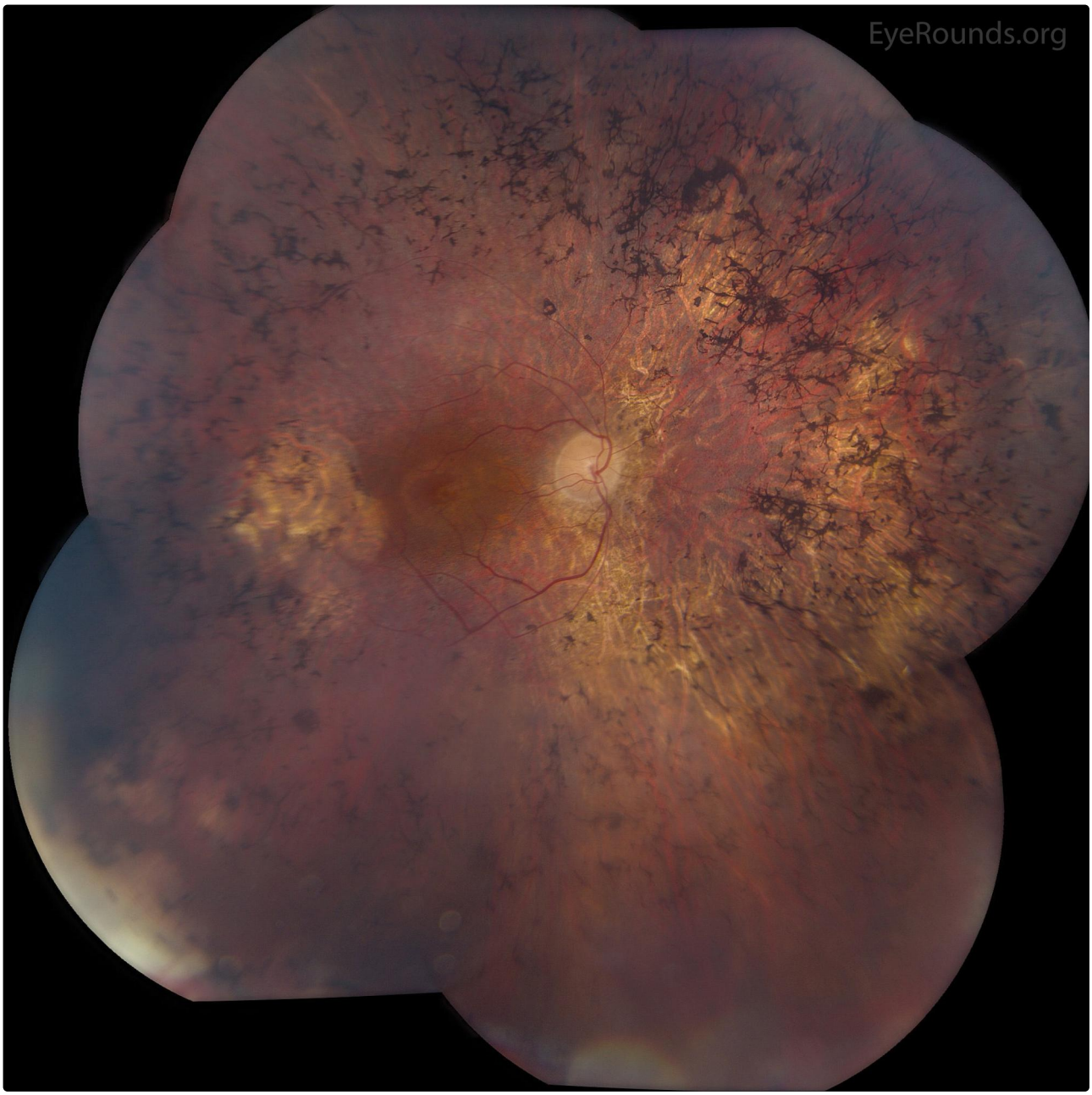
Photographer: Susan J. Wright

The montage color fundus photographs are from a 57-year-old male with molecularly confirmed retinitis pigmentosa (RP). He has a Pro347Ala change in the rhodopsin gene, which is consistent with autosomal dominant inheritance pattern. RP disease is rod-predominant photoreceptor degeneration, with late-stage atrophy of the cones and retinal pigment epithelium. It presents early in life with nyctalopia as a common first symptom. Visual field loss classically occurs in a ring-shaped scotoma with sparing of the central macula until late in the disease. Classic funduscopic findings include waxy-pallor of the optic disc, severe arteriolar attenuation, and bone-spicule-like pigmentation in the mid-peripheral retina. Routine dilated fundus examination and optical coherence tomography are important, as patients may develop cystoid macular edema, which may be respond to treatment with topical carbonic anhydrase inhibitors.



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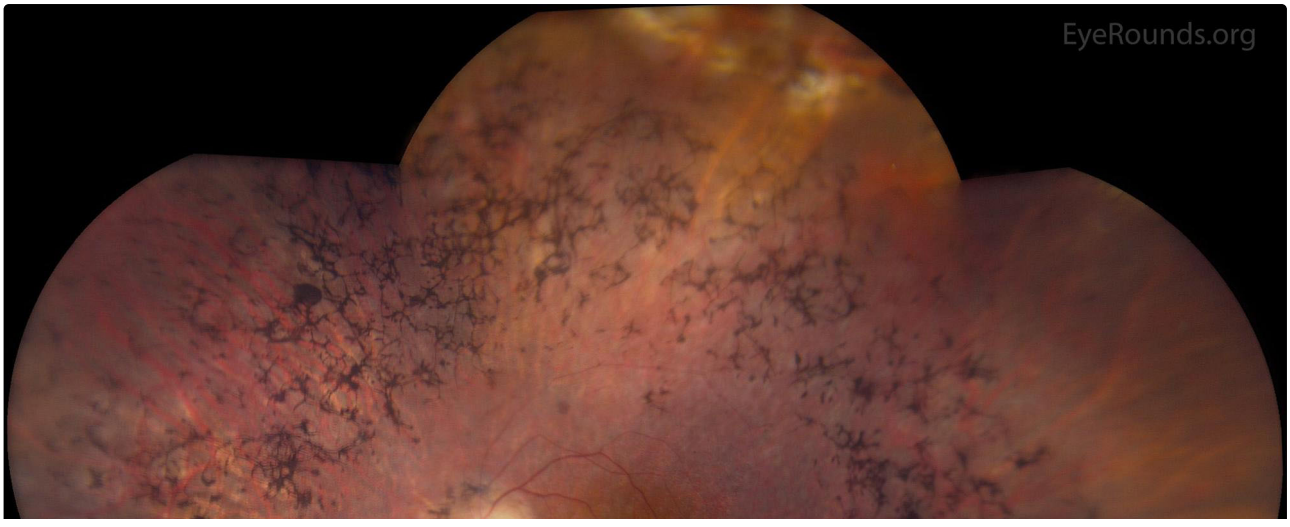

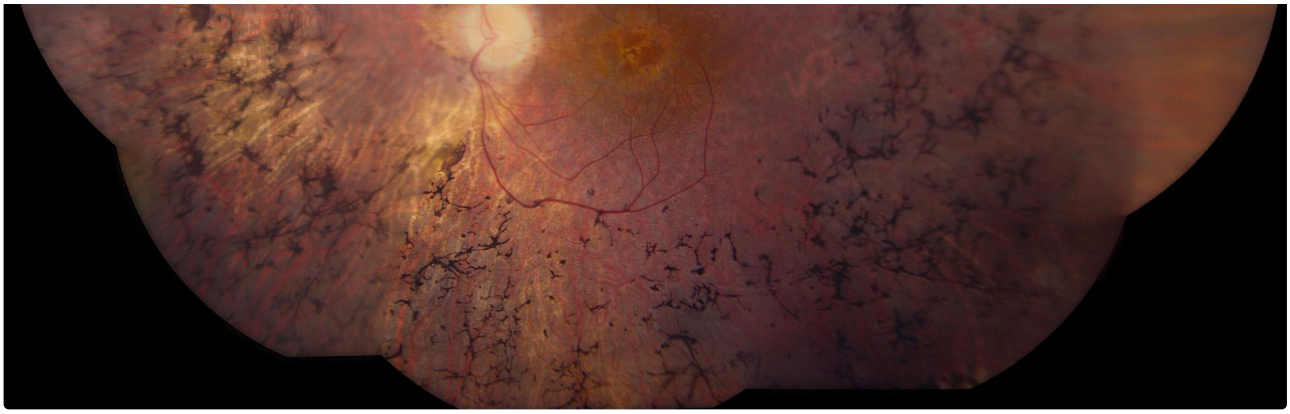

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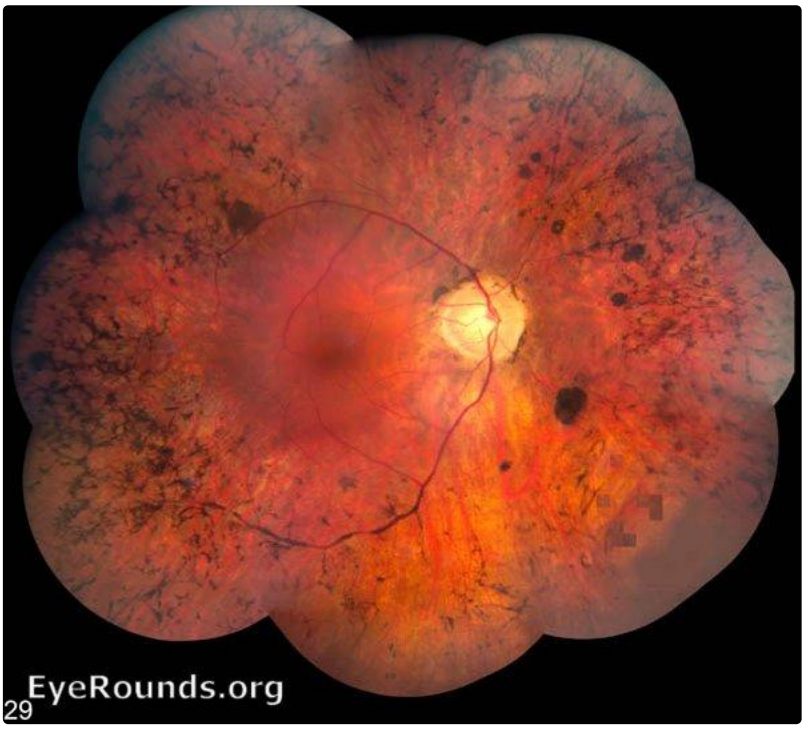
Category(ies): Retina

Contributor: Andrew Doan, MD, PhD



1. This is a montage color fundus photograph of the right eye in a patient with autosomal recessive RP. The photograph depicts waxy disc pallor, arteriolar attenuation, and bone-spicule like pigmentation of the peripheral retina (distributed along the microvascular pattern of the retina).

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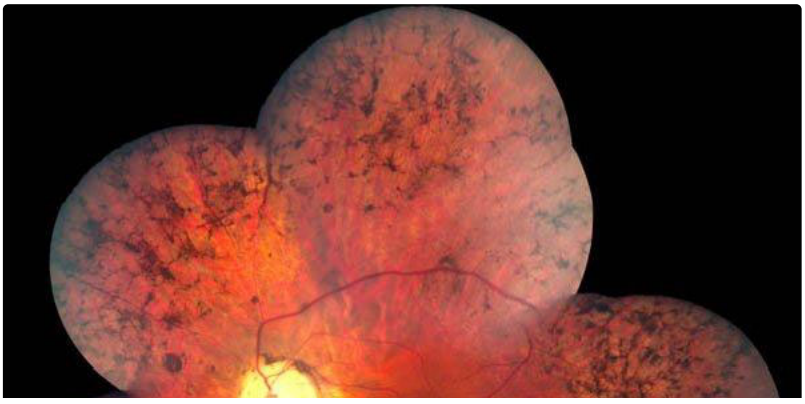

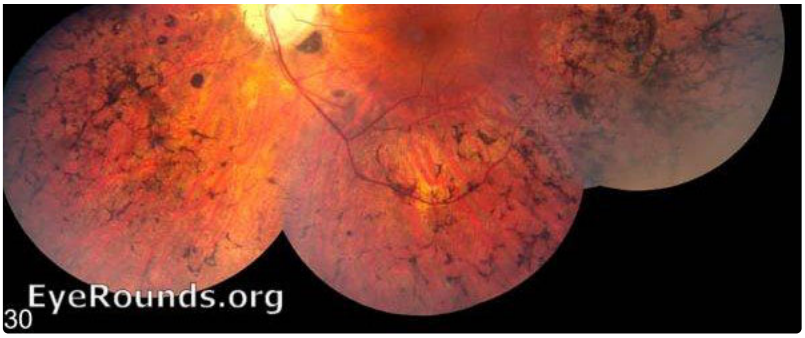

2. This is a montage color fundus photograph of the left eye in the same patient as above. Findings are similar to the right eye.

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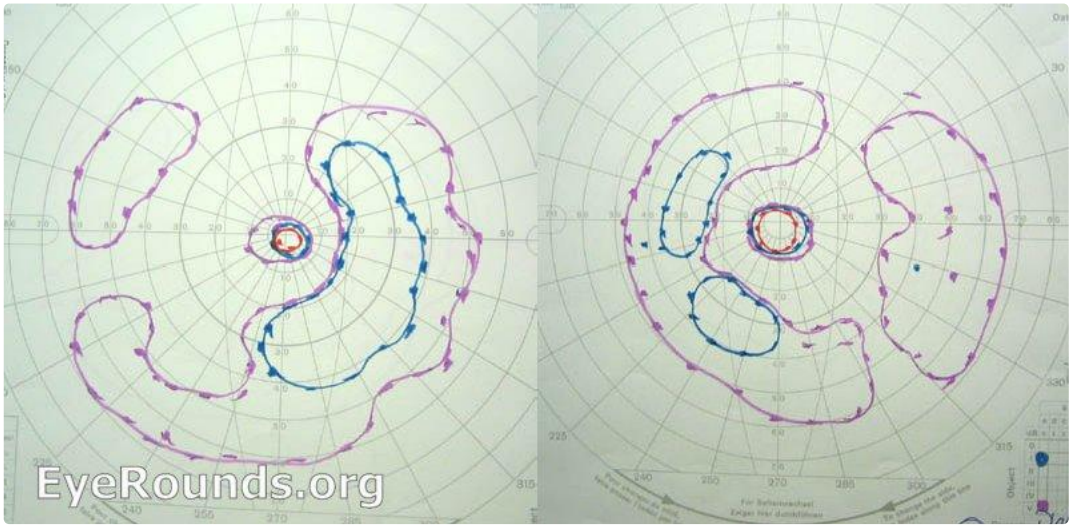

3. Goldmann visual perimetry of the right and left eye in the same patient as above demonstrating bilateral ring-pattern scotomas.

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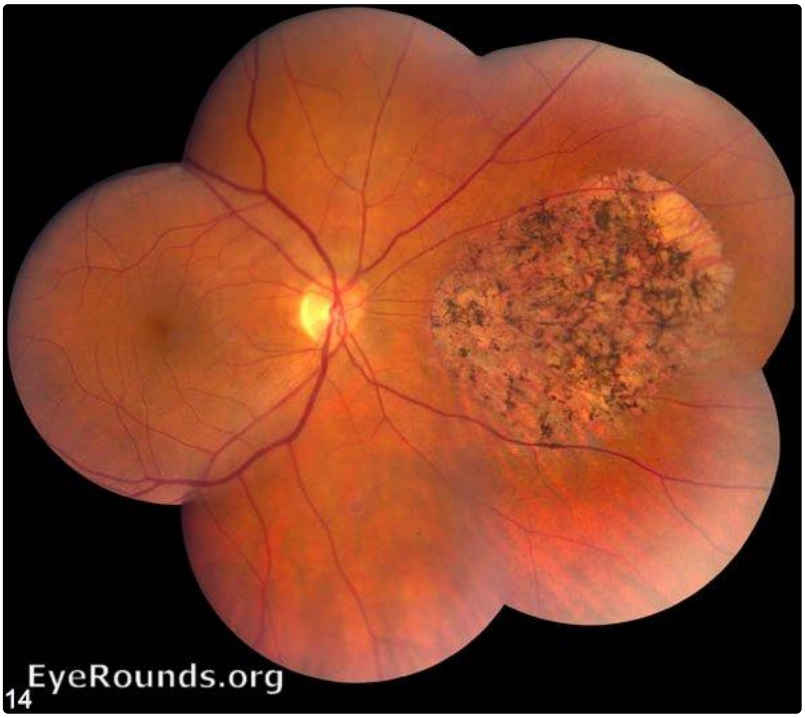

4. This is a montage color fundus photograph of the right eye in a female with x-linked RP in the carrier state. The photograph depicts unilateral, patchy pigmentation representing lyonization/x-inactivation. REFERENCE(S

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References:

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

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