Oculocutaneous albinism

Category(ies): Neuro-ophthalmology

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This 7-year-old girl presented with decreased vision and nystagmus and was diagnosed with oculocutaneous albinism (OCA).

OCA is typically inherited in an autosomal recessive fashion and is characterized by decreased melanin and subnormal vision since birth. Findings on exam include hypopigmentation of the skin, hair, and irides with diffuse iris transillumination defects on retroillumination, as seen in the photograph. Decreased acuity can be attributed to foveal hypoplasia which is apparent as an abnormally small or absent foveal depression as seen in this optical coherence tomography (OCT) render. Albinism can also be limited solely to the eyes in ocular albinism which most commonly has X-linked recessive inheritance.

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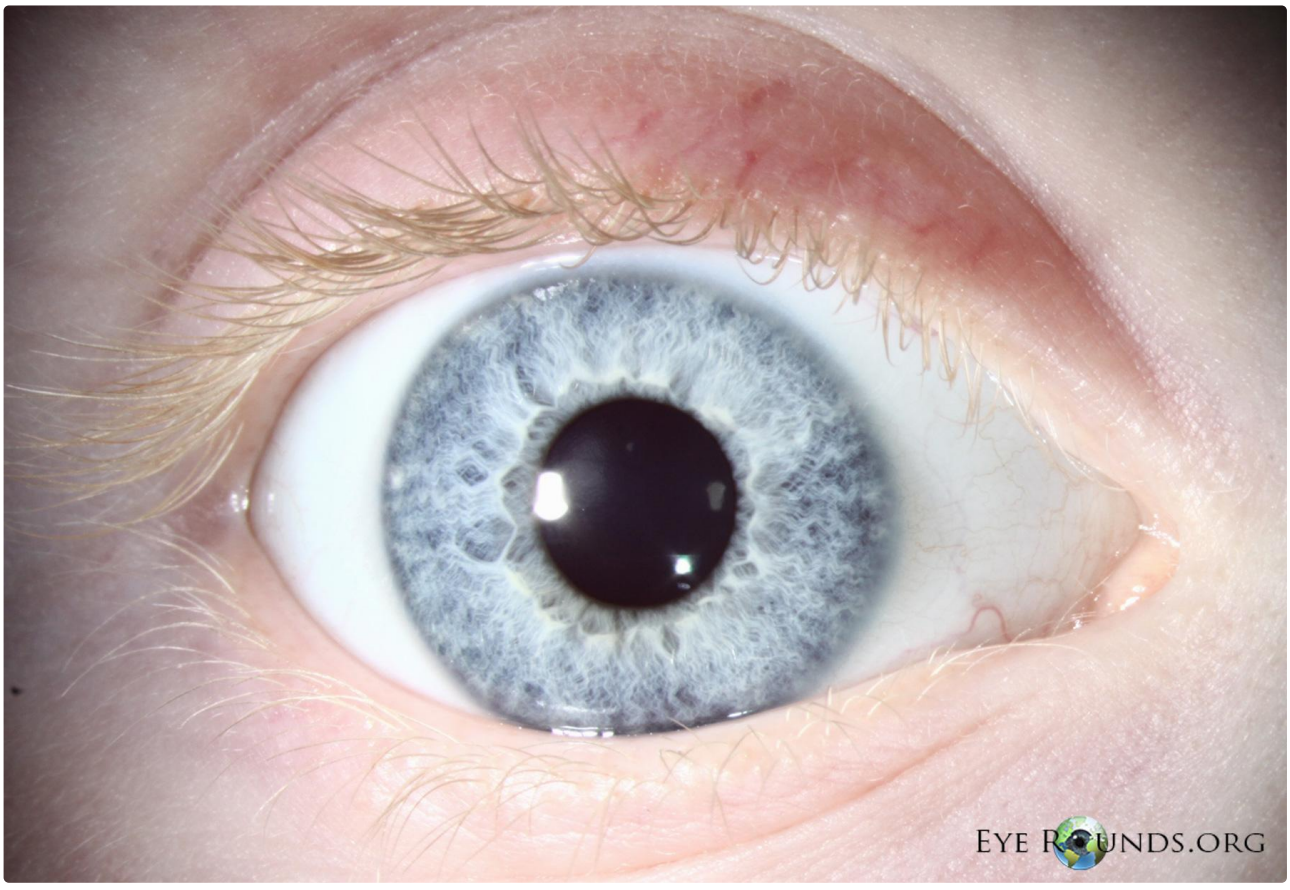

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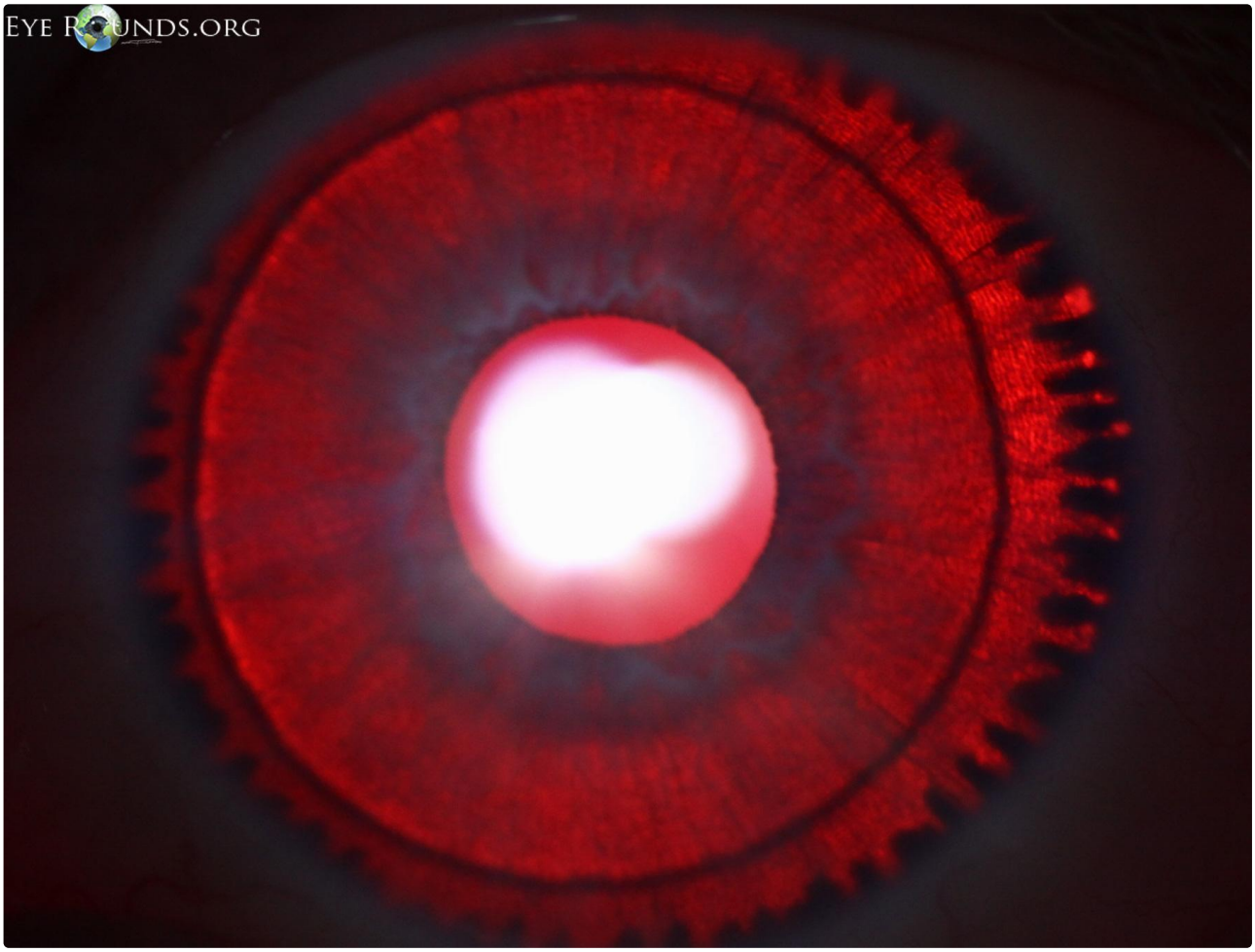

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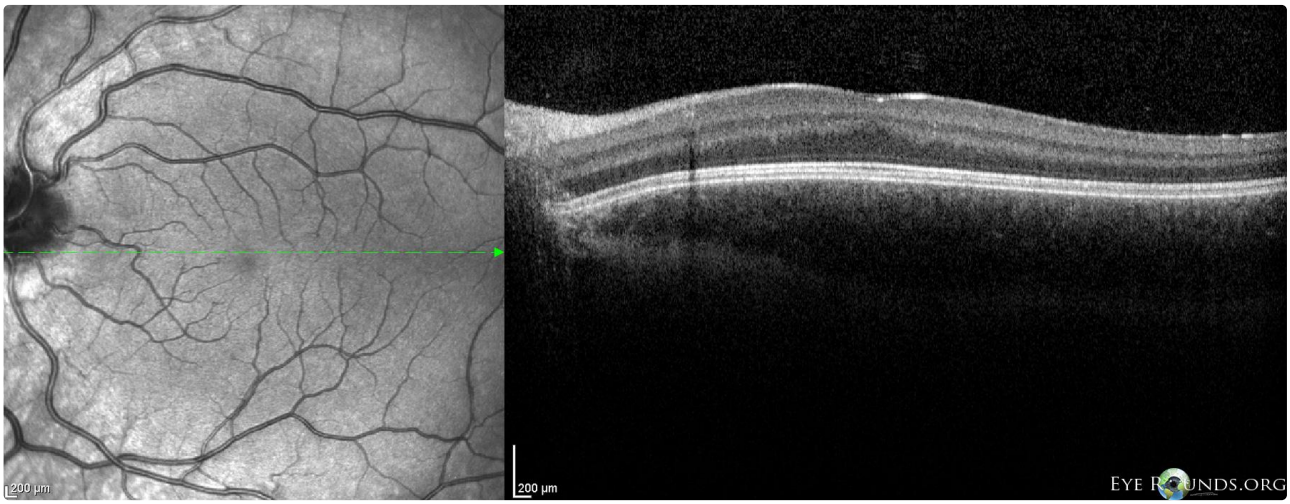

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