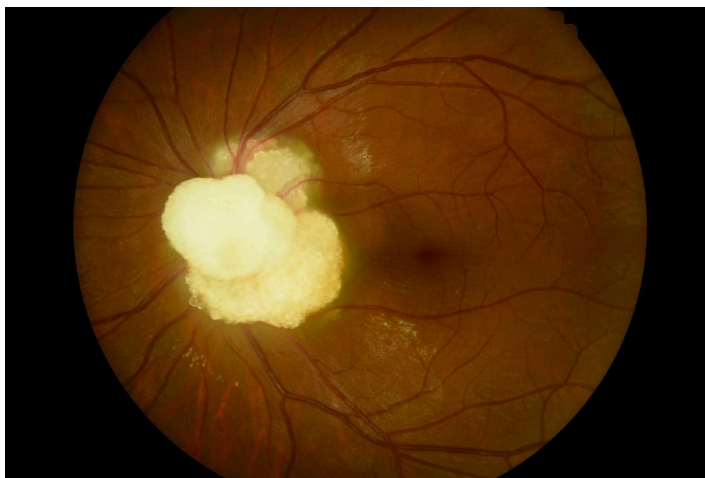


WHAT IS THIS MONTH'S MYSTERY CONDITION? Visit aao.org/eyenet to make your diagnosis in the comments.



LAST MONTH'S BLINK

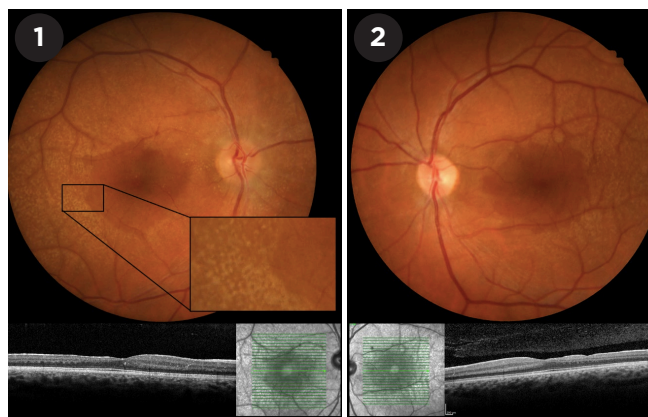
Retinal Findings in Alport Syndrome

A 20-year-old White man with Alport syndrome presented for his first-ever ocular evaluation. He had been diagnosed with Alport during childhood in the context of a positive family history, sensorineural deafness, and progressive kidney dysfunction. The patient had been under dialysis treatment for end-stage renal failure for two years before we saw him.

On examination, BCVA was 20/25 in both eyes. Anterior segment examination showed no evidence of anterior lenticonus or other abnormal findings.

However, fundus exam of both eyes showed foveal-sparing retinal flecks, associated with the retinal “lozenge” or “dull macular reflex” typical of Alport syndrome (Figs. 1,2). OCT showed symmetrical temporal macular thinning, also consistent with the disease.

Alport syndrome is a rare genetic disorder caused by mutations in three human genes in-



involved in type IV collagen biosynthesis. It commonly presents with characteristic retinal findings. The visual prognosis of these patients is favorable.

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