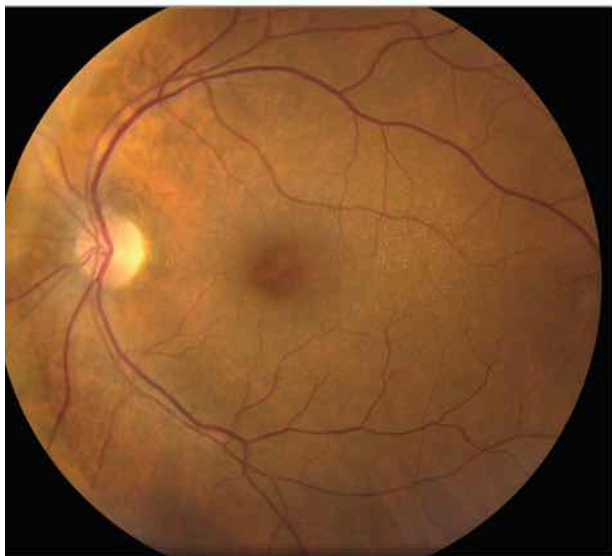


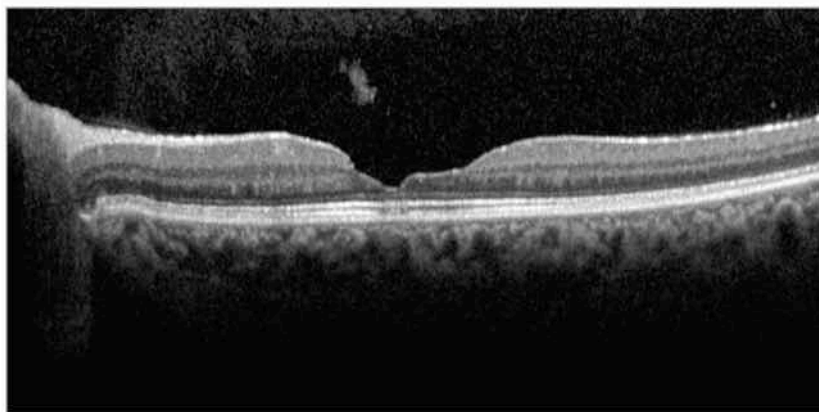
Staircase Lamellar Macular Hole in a Male Patient Aged 54 Years

Alessandro Marchese, MD^{1,2,3}; Michael Harrell Jr, MD¹; Manjot K. Gill, MD¹

Fundus photography



B Optical coherence tomography



A 54-year-old male patient presented for evaluation of a lamellar macular hole in his left eye. He had a full-thickness macular hole in his right eye that was operated on, 2 kidney transplants, and hearing loss from the age of 21 years. He denied a family history of ophthalmologic disorders, kidney disease, or hearing loss. His visual acuity was 20/40 OS. Fundus examination in the left eye showed temporal disc pallor and an abnormal macular reflex with superficial yellow retinal dots (Figure). Optical coherence tomography (OCT) revealed a lamellar macular hole in the left eye with a staircase appearance (Figure). The presence of kidney disease, hearing loss, and dot maculopathy with a staircase appearance on OCT strongly suggested Alport syndrome. Collagen alterations in basement membranes can lead to this maculopathy. Based on these findings, the patient received genetic testing leading to the diagnosis of X-linked Alport syndrome. Genetic testing showed a likely pathogenic variant in the *COL4A5* gene (c.2287G>C, p.Gly763Arg).

