Ophthalmic Images

Staircase Lamellar Macular Hole in a Male Patient Aged 54 Years

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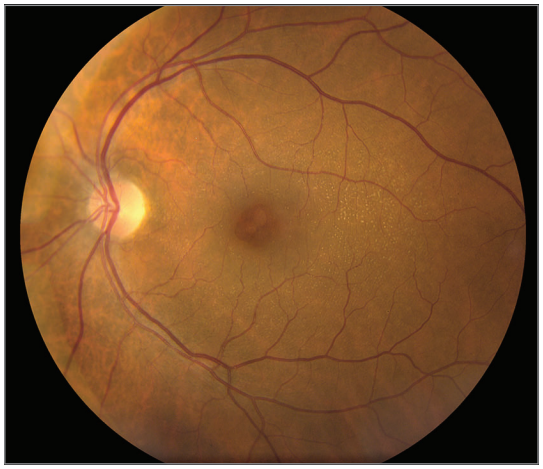
A Fundus photography



B Optical coherence tomography

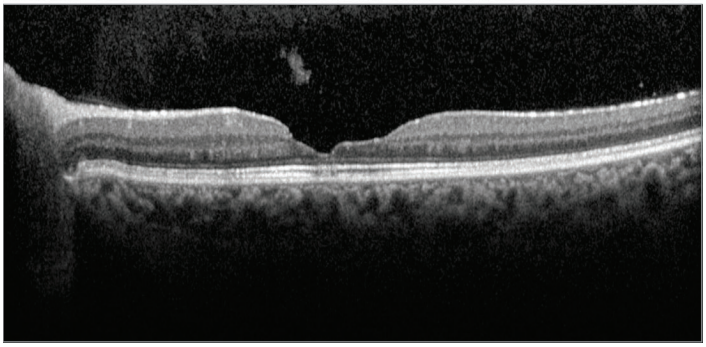


Figure. A, Color fundus photograph of the left eye shows a lamellar macular hole and multiple yellow dots scattered throughout the surface of the macula. B, Optical coherence tomography (confirms the lamellar macular hole; also, it reveals a staircase appearance of the fovea associated with highly reflective particles corresponding to the dot maculopathy seen in the color fundus photograph.

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**).

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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.[1,2] Collagen alterations in basement membranes can lead to this maculopathy.[1,2] 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).

ARTICLE INFORMATION

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Conflict of Interest Disclosures: Dr Marchese reported receiving personal fees from Kilabs Pharma. Dr Gill reported receiving grants from Research to Prevent Blindness and consultant fees from Genentech-Roche and Regeneron. No other disclosures were reported.

Additional Contributions: We thank the patient for granting permission to publish this information.

REFERENCES

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2. Cicinelli MV, Ritter M, Ghossein C, et al. The spectrum of internal limiting membrane disease in Alport syndrome: a multimodal imaging study. *Retina*. 2022;42(2):274-282. doi:10.1097/IAE.0000000000003295