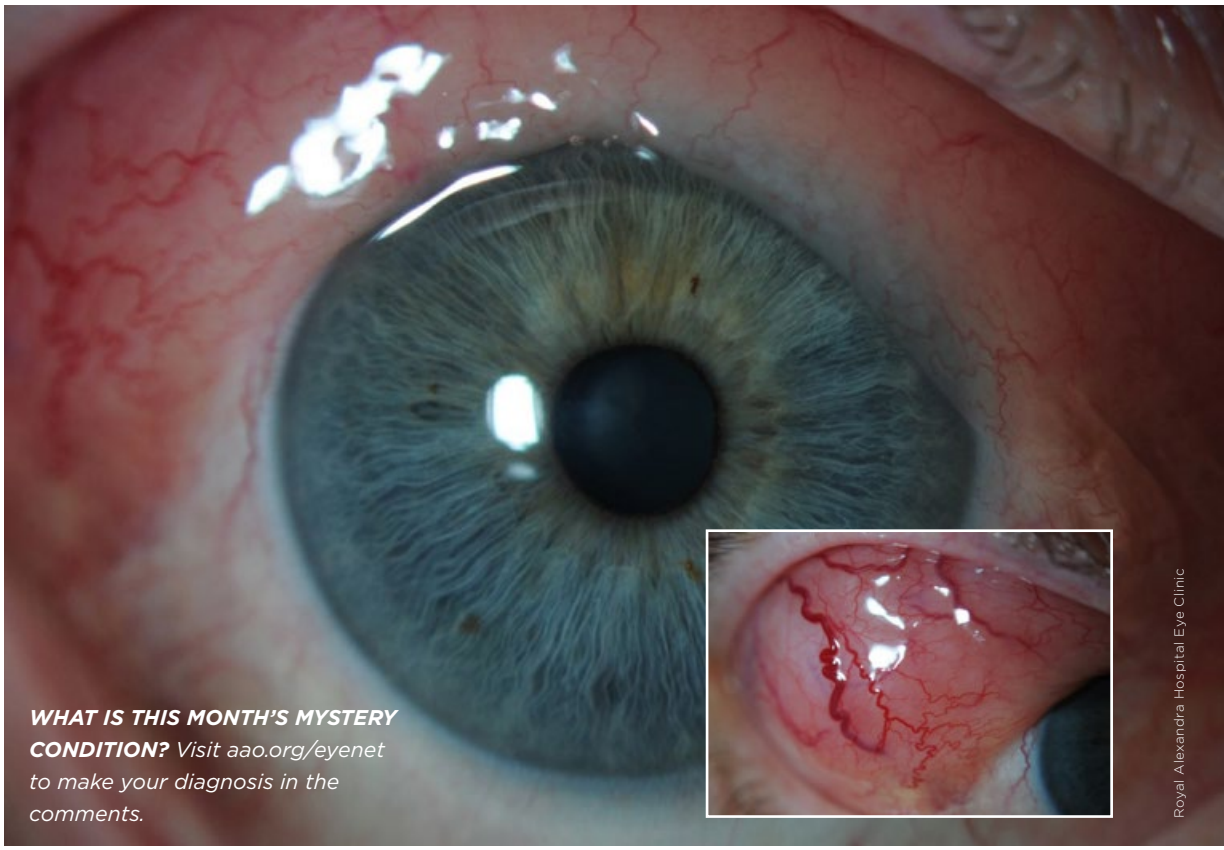
MYSTERY IMAGE

BLINK

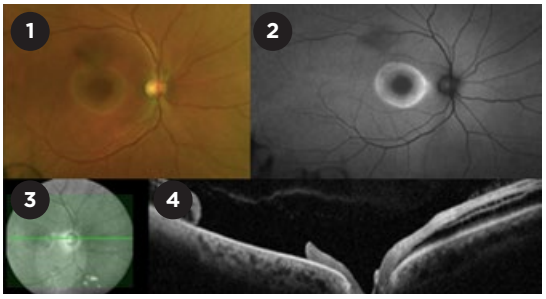


LAST MONTH'S BLINK

Giant Full-Thickness Macular Hole Associated With Alport Syndrome

A 49-year-old man with chronic poor vision in both eyes presented with worsening vision in his right eye. His past medical history was significant for glomerulonephritis, two kidney transplants, and sensorineural hearing loss. There was no history of trauma or ocular disease.

The patient's best-corrected visual acuity was 20/150 in his right eye and 20/60 in his left. Fundus examination revealed a giant full-thickness macular hole (FTMH) in the right eye (Fig. 1) and a partial-thickness macular hole in the left eye. Autofluorescence imaging of the right eye (Fig. 2) showed a typical bull's-eye appearance, with a central area of hypoautofluorescence and a surrounding rim of hyperautofluorescence. Optical coherence tomography of the right eye demonstrated a FTMH measuring 3,900 µm in diameter (Fig. 3) and retinoschisis involving the macula and the midperipheral retina (Fig. 4). Next-generation sequencing testing revealed a pathogenic *COL4A5* mutation consistent with Alport syndrome, a systemic disease that also affected his daughter.



Giant macular hole associated with Alport syndrome is thought to be caused by collagen abnormalities in the internal limiting membrane, Bruch membrane, or retinal pigment epithelium. Surgical treatment offers limited results.[1]

1 Miller JJ et al. *Retin Cases Brief Rep.* 2007;1(3):153-155.

WRITTEN BY **NATALIE HUANG, MD,** AND **SANDRA R. MONTEZUMA, MD.** PHOTOS BY **DREW MILLER.** ALL ARE AT UNIVERSITY OF MINNESOTA DEPARTMENT OF OPHTHALMOLOGY AND VISION NEUROSCIENCES, MINNEAPOLIS.