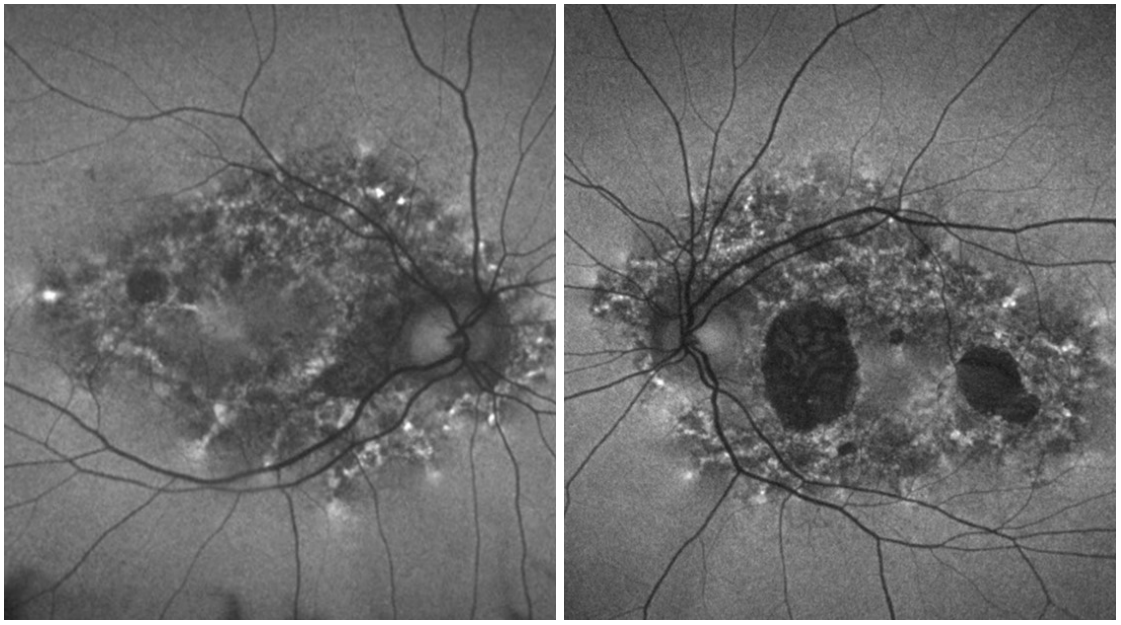
MYSTERY IMAGE

BLINK



George Henry, CRA, PBT (ASCP), Wheaton Eye Clinic, Wheaton, Ill.

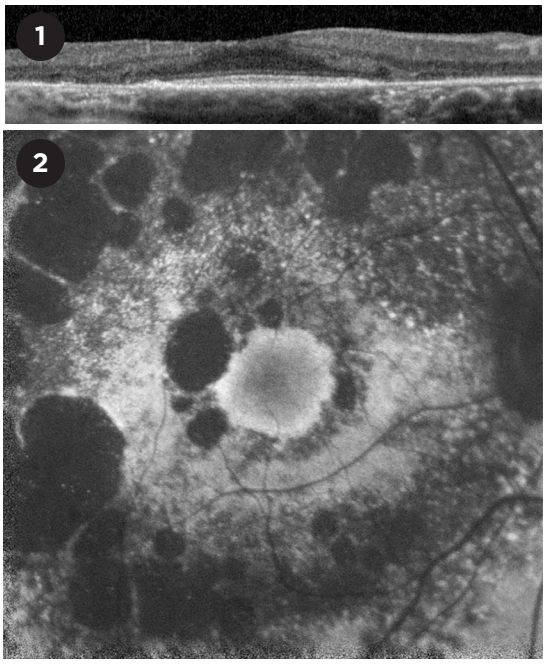
WHAT IS THIS MONTH'S MYSTERY CONDITION? *Visit aao.org/eyenet to make your diagnosis in the comments.*

LAST MONTH'S BLINK

NR2E3 Mutation

A 54-year-old man with a history of diabetes mellitus diagnosed at age 40 was referred for retinal findings on a routine exam. Visual acuity was 20/25 in each eye. Anterior segment exam revealed mild nuclear sclerosis of the lens. Dilated fundus exam demonstrated diffuse retinal pigment epithelium atrophy with pigment deposition and a bull's-eye pattern. OCT exhibited diffuse outer retinal atrophy, sparing the foveal region (Fig. 1). Fundus autofluorescence showed an array of lobules and specks of hypoautofluorescence arranged in concentric rings in the macula of both eyes (as shown in Fig. 2 of the right eye).

After medication toxicities were ruled out by the patient's history, the fundus autofluorescence findings helped to narrow down the differential diagnosis to mitochondrial conditions and certain retinitis pigmentosa variants. These unusual double concentric autofluorescence rings have been described with *NR2E3* mutations. Genetic testing was performed, confirming a heterozygous variant in *NR2E3*, consistent with autosomal dominant retinitis pigmentosa in this patient. Notably, autosomal recessive mutations in *NR2E3* have also been linked to enhanced S-cone syndrome, which tends to present with nyctalopia earlier in life.



WRITTEN BY **MICHAEL T. ANDREOLI, MD**, NORTHWESTERN MEDICINE, NAPERVILLE, ILL. PHOTOS BY **GEORGE HENRY, CRA, PBT (ASCP),** WHEATON EYE CLINIC, WHEATON, ILL.