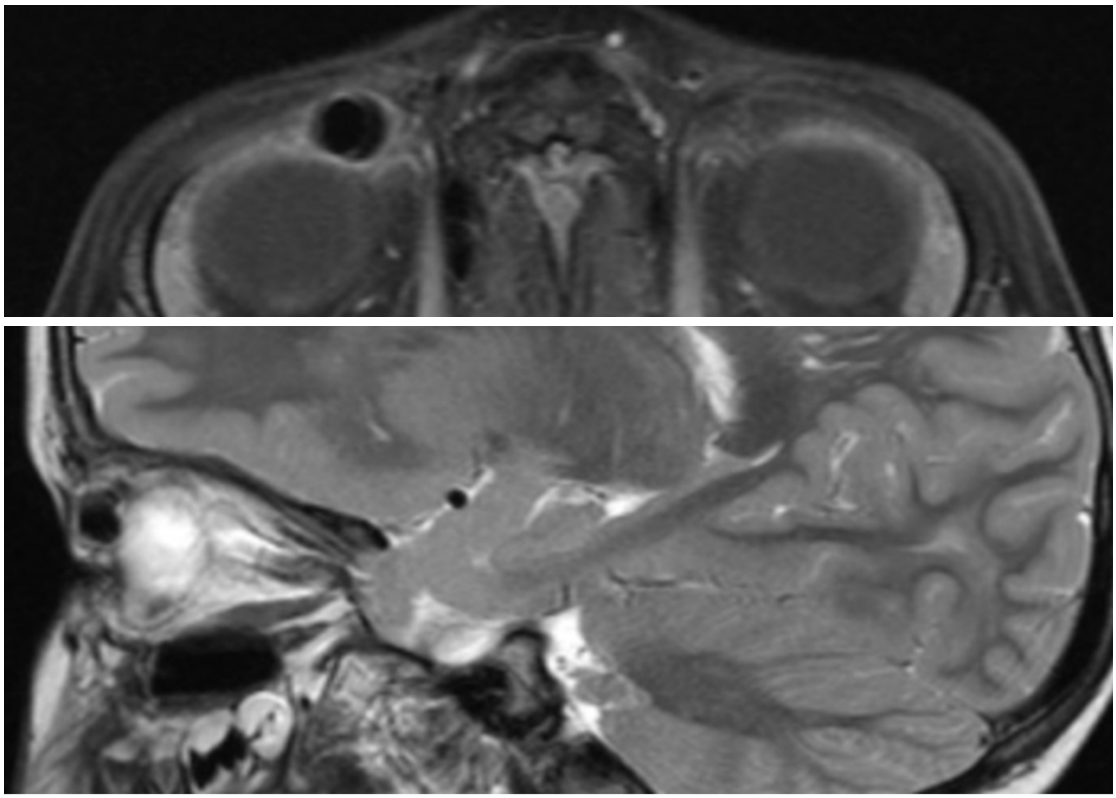
MYSTERY IMAGE

BLINK

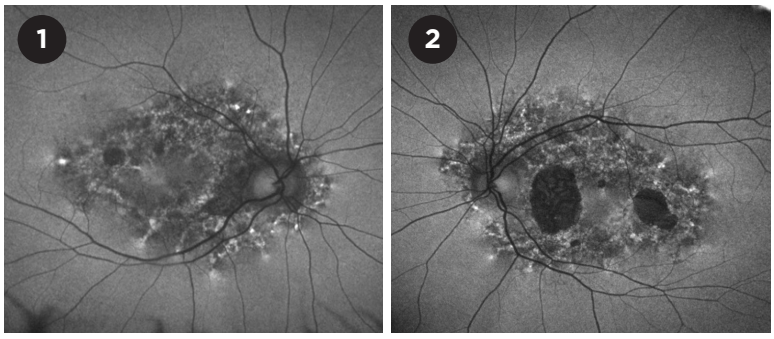


Melanie Daulton, MD, Northwestern University, Feinberg School of Medicine, Chicago.

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

LAST MONTH'S BLINK

MELAS Syndrome



A 63-year-old woman was referred for decreased vision. The patient had been diagnosed with multiple sclerosis (MS) at age 35 and diabetes mellitus at age 54; in addition, over the past five years, she had experienced multiple strokes and developed dementia. Her family history was significant for a sister who died at age 55 from MS, another sister who was blind from diabetic retinopathy, a daughter with MS, and a second daughter with diabetes and hearing loss. The patient's visual acuity was 20/40 in the right eye and 20/50 in the left. The fundus examination showed areas of atrophy and pigment hypertrophy in a reticulated pattern, as seen in the autofluorescent photos (Figs. 1, 2).

Genetic analysis was performed; it was positive for the mitochondrial mutation A>G at nucleotide position 3243 in both the patient and the daughter with diabetes and hearing loss. Because of heteroplasmy of the mutant mitochondrial DNA and other factors, phenotypic expression of the m3243 A>G mutation varies widely. Thus, while the mother's diagnosis is MELAS (Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-like episodes) syndrome, the daughter's diagnosis would be maternally inherited diabetes and deafness (MIDD). Genetic counseling was recommended for family members.

WRITTEN BY **MARK J. DAILY, MD.** PHOTO BY **GEORGE E. HENRY, CRA, P (ASCP).** BOTH ARE AT WHEATON EYE CLINIC, WHEATON, ILL.