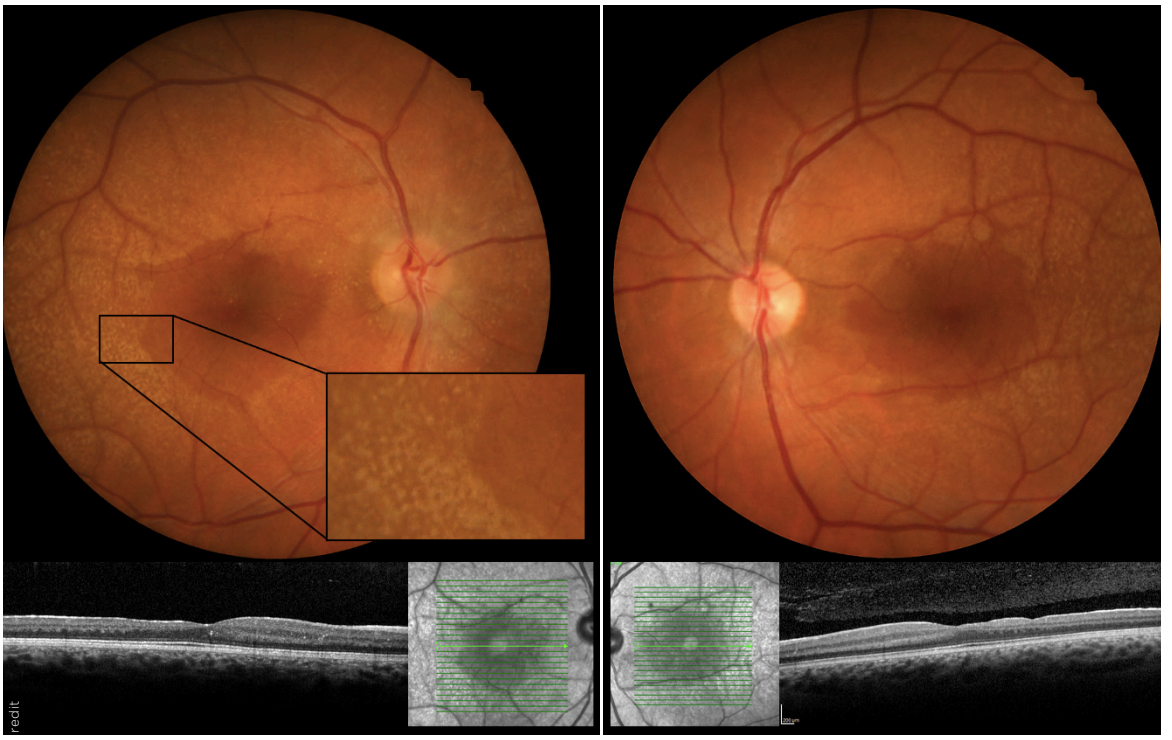


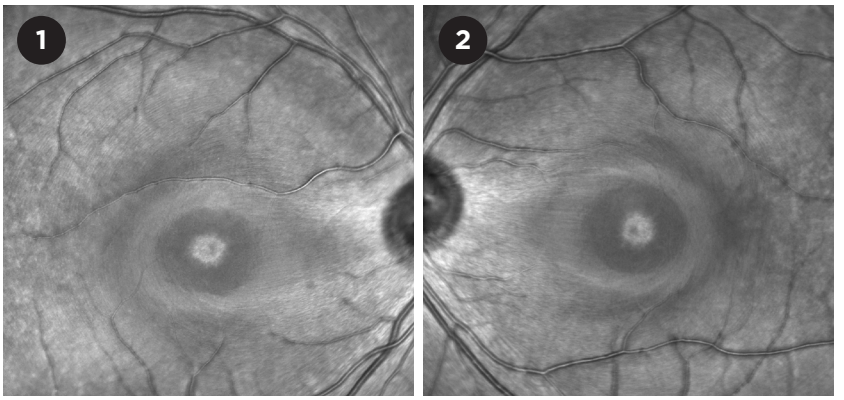
Inês Nicho, Hospital Prof. Doutor Fernando Fonseca, Lisbon, Portugal.

reedit

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

LAST MONTH'S BLINK

Sialidosis Type 1



An asymptomatic 14-year-old boy was referred for a second opinion of cherry-red spots in his maculae. The best-corrected visual acuity was 20/20 in both eyes. The slit-lamp exam revealed snowflake cataracts, and the fundus exam found perifoveal graying with a cherry-red spot in both maculae. OCT showed deposits in the ganglion cell layer, and no leakage was found with fluorescein angiogram. Fundus autofluorescence (Figs. 1, 2) revealed a bull's-eye appearance to the maculae with hypoautofluorescence surrounding a hyperautofluorescent center.

Genetic testing through Invitae Comprehensive Lysosomal Storage Disorders Panel revealed a pathogenic variant in *NEU1* (neuraminidase 1) and a variant of unknown significance in *SMPD1* (sphingomyelin phosphodiesterase 1).

The patient was diagnosed with sialidosis type 1. This gene mutation causes a lysosomal storage disease that is inherited as an autosomal recessive trait.

The patient's family members came in for genetic testing and imaging. His 18-year-old brother also was diagnosed with sialidosis type 1; his father was diagnosed with MacTel (macular telangiectasia); and his mother and sister were not found to have any gene mutations.

WRITTEN AND PHOTOGRAPHED BY **BECKY WEEKS, BS, CRA, OCT-C,** MORAN EYE CENTER, SALT LAKE CITY.