

# WAARDENBURG SYNDROME



A unique presentation leads to a rare diagnosis.

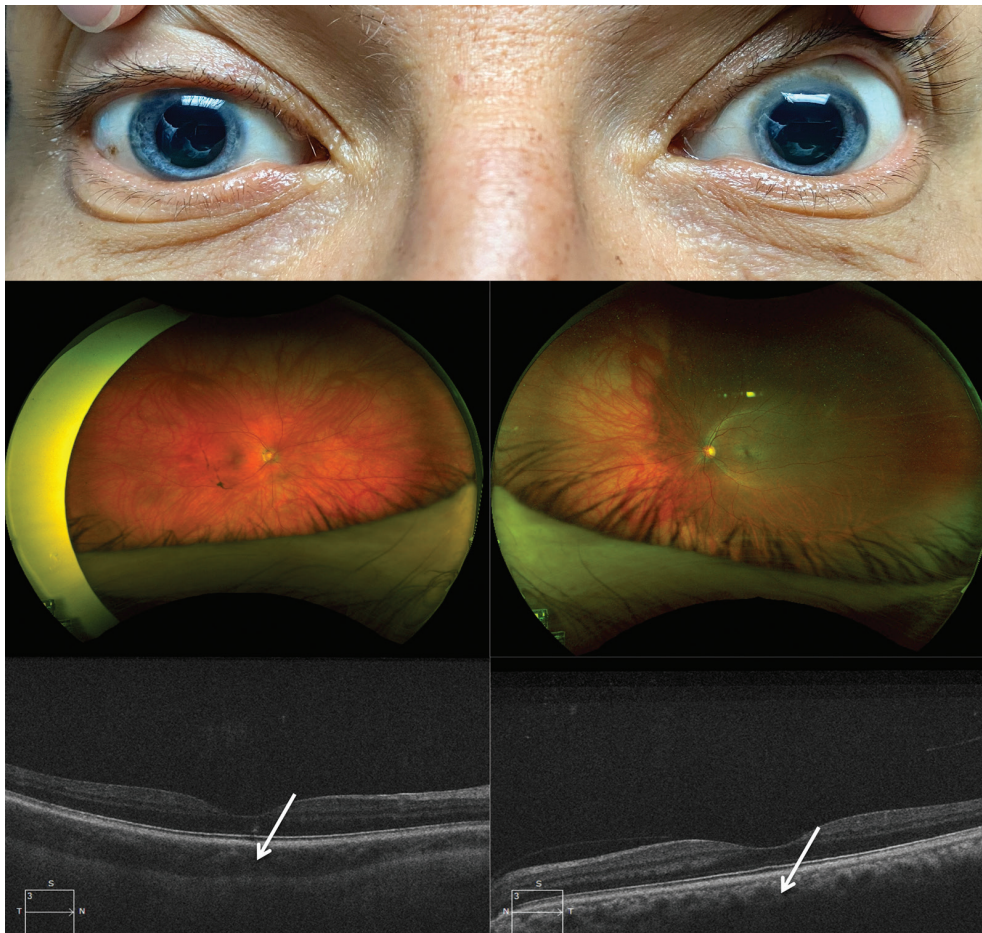
BY KUNYONG XU, MD, MHSC; DAVID R.P. ALMEIDA, MD, MBA, PHD; AND ERIC K. CHIN, MD

**A** 47-year-old Hispanic man presented with congenital iris heterochromia, white forelock, and bilateral hearing loss. VA was 20/25 in both eyes.

The patient has blue eyes, and iris hypopigmentation was noted in both eyes with focal areas of pigmentation superotemporally in the left eye (Figure, top). Hypopigmentation of the retina and choroid in both eyes was observed, and relative temporal hyperpigmentation was observed in the left eye only (Figure, middle). OCT of

the macula showed relative choroidal thinning in the right eye compared with the left eye (Figure, bottom, white arrows).

There was no history of malformation of upper extremities or Hirschsprung disease. External examination revealed no telecanthus, synophrys, or patches of skin depigmentation. There was no tubular nose and no small nasal alae. The patient was diagnosed with Waardenburg syndrome based on the findings and history. ■



**DAVID R.P. ALMEIDA, MD, MBA, PHD**

- Vitreoretinal Surgeon, Erie Retinal Surgery, Erie, Pennsylvania
- drpa@pm.me;
- Twitter: @davidalmeidaMD
- Financial disclosure: Advisory Board, Consultant, Speaker, (Alcon, Allergan, Bayer, Genentech, Novartis, Regeneron); Cofounder, Equity Holder (Citrus Therapeutics)

**ERIC K. CHIN, MD**

- Private practice, Retina Consultants of Southern California, Redlands, California
- Assistant Professor, Loma Linda University; Loma Linda Veterans Affairs Hospital
- chin.eric@gmail.com
- Financial disclosure: Research Investigator (Allergan, Chengdu Kanghong Biotechnology, Genentech, Novartis, Senju Pharmaceuticals), Cofounder, Equity Holder (Citrus Therapeutics)

**KUNYONG XU, MD, MHSC**

- Department of Ophthalmology and Visual Sciences, University of Arizona, Tucson, Arizona
- Financial disclosure: None