WAARDENBURG SYNDROME

A unique presentation leads to a rare diagnosis.

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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.

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