Teaching NeuroImages: Waardenburg syndrome type 2

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Figure 1 Vivid blue eyes

Bilateral isohypochromia iridis (vivid blue eyes) in our patient.

A 5-year-old boy presented with bilateral congenital sensorineural hearing loss and bilateral isohypochromia iridis without dystopia canthorum. MRI showed a shortened posterior semicircular canal (figures 1 and 2) compatible with Waardenburg syndrome (WS).

WS is a rare genodermatosis that affects 1:40,000 children and is caused by abnormal migration of melanoblasts from the neuroectoderm. There are 4 types of WS (1 and 3, autosomal dominant; 2 and 4, dominant or recessive). Type 2 differs from type 1 due to the absence of dystopia canthorum. Type 3 is associated with musculoskeletal abnormalities and type 4 with Hirschsprung disease.^{1,2}

Author contributions

M. Rosa Júnior: design or conceptualization of the study, analysis or interpretation of the data, drafting or revising the manuscript for intellectual content. L.M. Santana: analysis or interpretation of the data. B.F. Ramos: analysis or interpretation of the data, drafting or revising the manuscript for intellectual content. H.F. Ramos: analysis or interpretation of the data, drafting or revising the manuscript for intellectual content.

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Disclosure

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Figure 2 MRI in Waardenburg syndrome

(A–C) Axial T2-weighted imaging (T2WI) shows normal superior semicircular canal (SC) (white arrows), normal lateral SC (yellow arrows), and shortened and thick posterior SC (red arrows). (D) Sagittal T2WI and (E) 3D posterior view show normal superior SC (white arrow) and abnormal posterior SC (red arrow).

References

1. Read AP, Newton VE. Waardenburg syndrome. *J Med Genet* 1997;34:656–665.
2. Soni CR, Kumar G. Child Neurology: a patient with dissimilar eye color and deafness. *Neurology* 2010;74:e25–e26.

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