Teaching NeuroImages: Trigeminal Ganglia Hypoplasia as Imaging Clue for the Diagnosis of Gómez-López-Hernández Syndrome

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Figure 1 Brachycephaly, Midface Hypoplasia, Strabismus, and Left Unilateral Parietal Alopecia in Gómez-López-Hernández Syndrome

Patient with Gómez-López-Hernández syndrome. Frontal and lateral view of the face shows brachycephaly, midface hypoplasia, strabismus, and left unilateral parietal alopecia.

A 42-year-old woman presented with ataxia and numbness in her face since childhood. Her parents were nonconsanguineous. Examination showed ataxia, alopecia, absent corneal reflex, and hypoesthesia in right trigeminal territory (figure 1). Brain MRI disclosed rhombencephalosynapsis and trigeminal nerve hypoplasia (figure 2). Exome sequencing excluded other genetic diseases. Gómez-López-Hernández syndrome (GLHS) was diagnosed.

GLHS is a rare and maybe underdiagnosed neurocutaneous disorder (with few cases described) characterized by ataxia, alopecia, trigeminal anesthesia, rhombencephalosynapsis, and brachyturrycephaly. Genetics of GLHS is unknown.[1] Brain imaging includes rhombencephalosynapsis, which may occur in other syndromes. Trigeminal ganglia hypoplasia is a neuroimaging clue for the diagnosis of GLHS.[2]

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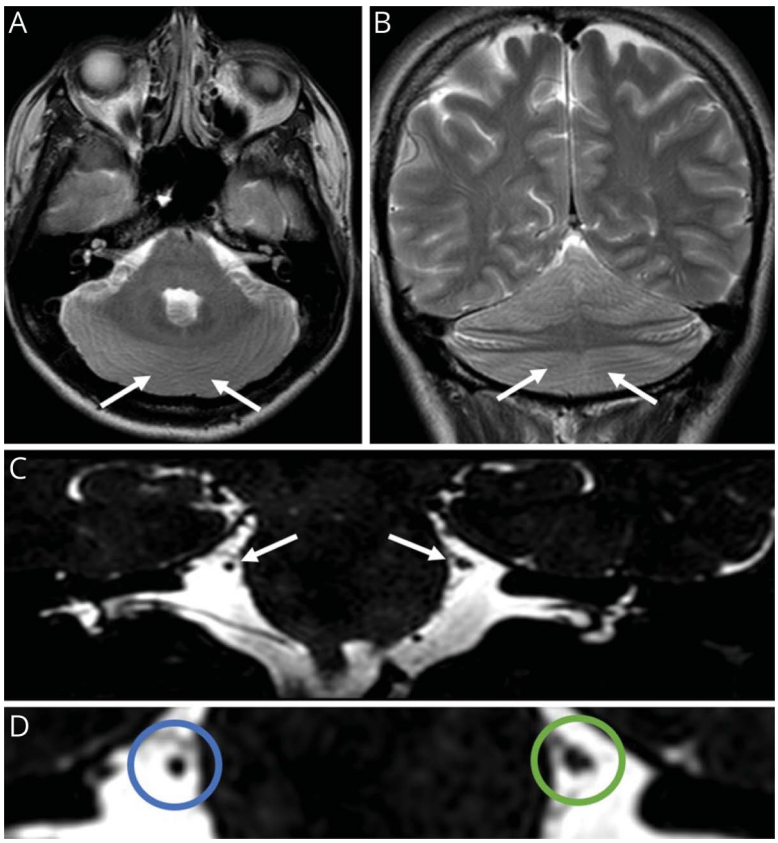
Figure 2 MRI Shows Rhombencephalosynapsis and Hypoplasia of the Right Trigeminal Nerve in Gómez-López-Hernández Syndrome



Axial (A) and coronal (B) T2-weighted brain MRI shows rhombencephalosynapsis, characterized by absence of vermis, with fusion of the cerebellar hemispheres (arrows). (C) Coronal FIESTA shows hypoplasia of the right trigeminal nerve (arrows). (D) Expanded imaging highlights hypoplasia of the right trigeminal nerve (blue circle) that is thinner than the normal contralateral nerve (green circle).

Disclosure

The authors declare no disclosures relevant to the manuscript. Go to Neurology.org/N for full disclosures.

Appendix Authors

Name	Institution	Contribution
Eduardo Perrone, MD	Universidade Federal de São Paulo, Brazil	Designed and conceptualized the case report, collected the data, wrote the first draft, revised the manuscript
Stênio Burlin, MD	Universidade Federal de São Paulo, Brazil	Designed and conceptualized the case report, collected the data, revised the manuscript
Vânia D'Almeida, PhD	Universidade Federal de São Paulo, Brazil	Designed and conceptualized the case report, revised the manuscript
Ana Beatriz Alvarez Perez, PhD	Universidade Federal de São Paulo, Brazil	Designed and conceptualized the case report, revised the manuscript
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Appendix *(continued)*

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Victor Rebelo Procaci, MD	Universidade Federal de São Paulo, Brazil	Designed and conceptualized the case report, collected the data, wrote the first draft, revised the manuscript
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Orlando G. Barsottini, MD, PhD	Universidade Federal de São Paulo, Brazil	Designed and conceptualized the case report, revised the manuscript
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