Mystery Case: Bilateral alopecia as clue to diagnosis of Gomez-Lopez-Hernandez syndrome in a 38-year-old man

Cornelius Kronlage, MD, and Daniel G. Healy, MD

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Correspondence
Dr. Kronlage
cornelius.kronlage@usb.ch

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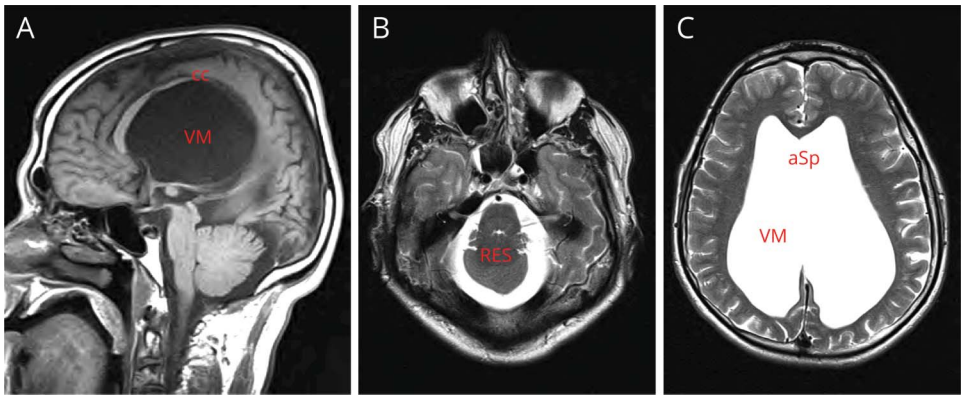
Figure 1 Brain imaging findings in Gomez-Lopez-Hernandez syndrome (GLHS)



(A) Sagittal T1-weighted and (B, C) axial T2-weighted MRI show hypoplasia of brainstem and cerebellum with absence of the vermis—signifying rhombencephalosynapsis (RES)—and marked ventriculomegaly (VM). Other findings common in GLHS include absent septum pellucidum (aSp) and dysplastic corpus callosum (cc).

A 38-year-old man with a diagnosis of cerebral palsy had MRI for headache. This showed hydrocephalus with aqueductal stenosis and a hypoplastic cerebellum (figure 1). On detailed examination, bilateral triangular patches of alopecia (figure 2) stood out. Subsequently, absent corneal reflexes were found, while careful review of the imaging revealed rhombencephalosynapsis (RES).

The triad of alopecia, trigeminal anesthesia, and RES defines Gomez-Lopez-Hernandez syndrome (GLHS), a rare congenital malformation syndrome of unknown etiology. GLHS is part of a spectrum of RES-associated malformations[1] that may account for as many as 9% of congenital aqueductal stenoses.[2]

Hair abnormalities are also distinctive in other rare neurologic diagnoses, for example, cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy (CARASIL) (premature alopecia), Menkes syndrome (curly, brittle hair; pili torti), Woodhouse-Sakati syndrome (premature, progressive alopecia), and giant axonal neuropathy (curly, "kinky" hair).

Author contributions

C. Kronlage assisted in the patient's clinical assessment, drafted and revised the manuscript, and created the figures. D. G. Healy performed the patient's clinical assessment and revised the manuscript and figures.

From the Department of Neurology (C.K.), University Hospital Basel, Switzerland; and Department of Neurology (C.K., D.G.H.), Beaumont Hospital, Dublin, Ireland.

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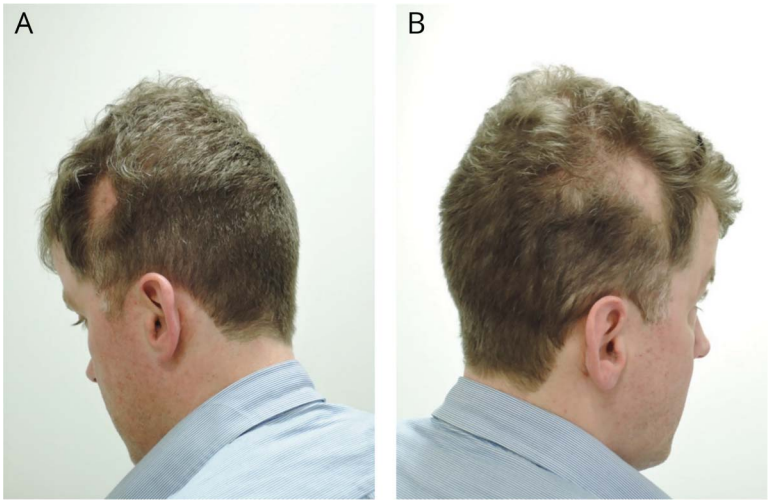
Figure 2 Hair abnormalities in Gomez-Lopez-Hernandez syndrome



(A, B) Bilateral triangular alopecia on the parieto-occipital scalp.

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Disclosure

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

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Mystery Case Responses

The Mystery Case series was initiated by the *Neurology*® Resident & Fellow Section to develop the clinical reasoning skills of trainees. Residency programs, medical student preceptors, and individuals were invited to use this Mystery Case as an educational tool. Responses to multiple choice questions formulated using this case were solicited through a group email sent to the American Academy of Neurology Consortium of Neurology Residents and Fellows and through social media. We received 129 responses. The majority of respondents (73%) had been in practice for 1–4 years; 68% were residents or fellows while 23% were faculty/board-certified physicians; the remainder were medical students or advanced practice providers. 65% resided outside the United States. A wide range of practice settings were represented.

The 38-year-old patient with a historic diagnosis of cerebral palsy underwent an MRI brain for investigation of headache. On reviewing the imaging, most respondents correctly identified hypoplasia of the cerebellar vermis and corpus callosum (63%) and ventriculomegaly (78%). However, only 36% identified brainstem hypoplasia. The most common incorrect answer was hydrancephaly (24%), a congenital brain defect in which the cerebral hemispheres are replaced by fluid-filled cavities, usually presenting in infancy with macrocephaly.[1]

Of the listed neurologic conditions, GLHS (40%), Vogt-Koyanagi-Harada syndrome (28%), and CARASIL (19%) were correctly identified as being associated with alopecia. The most common incorrect answers were zinc toxicity (44%; zinc deficiency can cause alopecia), phenytoin use (36%; associated with hirsutism), and Menkes syndrome (33%; a congenital X-linked disorder causing copper deficiency and associated with "kinky" hair[2]).

GLHS was correctly identified as being the triad of alopecia, trigeminal anesthesia, and RES by most respondents (53%). RES is the term given to the congenital cerebellar abnormalities seen in GLHS and other disorders, which are associated with cerebellar signs, cerebral palsy, and intellectual developmental delay.[3] The most common incorrect answer was Vogt-Koyanagi-Harada syndrome (12%), a multisystem disease caused by melanocyte inflammation and more common in darker skinned ethnicities. Features include chronic panuveitis, vitiligo, alopecia, dysacousia, meningeal irritation, and poliosis.[4]

This case highlights the characteristic features of a rare syndrome, recognition of which offers some diagnostic specificity for those labeled as having "cerebral palsy." Several neurologic disorders are associated with skin and hair abnormalities, and their appreciation can narrow differential diagnoses and aid identification of treatable causes.

Robert Hurford, MSc, MRCP (UK)
Nuffield Department of Clinical Neurosciences, University of Oxford

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