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Partial Rhombencephalosynapsis Presenting in an Adult with Cerebello-Trigeminal-Dermal Dysplasia

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Gomez-Lopez-Hernandez syndrome (GLHS), also known as cerebello-trigeminal-dermal dysplasia, is a neurocutaneous disorder typically presenting in childhood. GLHS is characterized by rhombencephalosynapsis (RES) and partial alopecia, with or without trigeminal anesthesia. We describe a rare case of GLHS in a paucisymptomatic adult who presented with new-onset seizure-like activity.

Magnetic resonance imaging revealed partial midline fusion of the cerebellar hemispheres, incomplete development of vermis, and slight medialization of the dentate nuclei: all consistent with the diagnosis of RES. Radiographic evidence combined with partial alopecia, truncal ataxia, and muscular hypotonia are suggestive GLHS diagnosis.

Our report not only highlights the importance of maintaining GLHS on the differential for newonset seizure-like activity, but also demonstrates how patients with GLHS may be minimally symptomatic and diagnosed in adulthood.

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