**Opinion**
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# Cerebello-trigeminal-dermal Dysplasia Syndrome

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## Opinion

Cerebello-trigemino-dermal dysplasia is a rare neurocutaneous syndrome of craniosynostosis, ataxia, trigeminal anesthesia, scalp alopecia, cerebellar anomaly, midface hypoplasia, corneal opacities, apparently low-set ears, mental retardation, and short stature. It seems to be a sporadic condition but little is known about its cause and pathogenesis in the few cases reported so far. We present three new unrelated patients and magnetic resonance images of the central nervous system, and review the four cases reported previously. We think that this is not such a rare condition, and that it is underdiagnosed. GLHS is an uncommon neurocutaneous syndrome, possibly a sporadic condition that is underdiagnosed. Due to the new imaging and genetic technologies pre and post-natal, today it is possible to achieve a better and more accurate diagnosis of hydrocephalus with a genetic origin, in which the high suspicion of teams of clinical specialists is essential. Without accurate diagnosis, we cannot access to a long-term prognosis, prevention of aggregate morbidity or an adequate genetic counseling, which are required in today's pediatrics.

Gomez Lopez Hernandez syndrome (GLHS) is a rare condition characterized by partial scalp baldness, numbness of the face, eyes, sinuses, and mouth and a brain abnormality called rhombencephalosynapsis. Additional symptoms vary but may include distinctive facial features, intellectual disability or other neurological problems, and craniofacial abnormalities. The cause of GLHS is poorly understood. It may be genetic, possibly with autosomal recessive inheritance. Treatment depends on the signs and symptoms present in each person. Hydrocephalus is defined as an active distension of the ventricular system, of the brain resulting from inadequate passage of cerebrospinal fluid from its point of production with the cerebral ventricles to its point of absorption into the systemic circulation. The prevalence varies from 0.5 to 3.2 per 1,000 new borns, whether or not hydrocephalus following intraperiventricular hemorrhage is included.

Causes of hydrocephalus can be classified according to the main clinical symptoms. There are classifications according to the onset, localization of lesions, pressure dynamics of CSF, non-genetic causes. Genetic causes of hydrocephalus are classified as isolated hydrocephalus or syndromic hydrocephalus. Genetic hydrocephalus must be investigated con sidering recurrence and family aggregation risks. Among SH, the Gómez-López-Hernández syndrome (GLHS) or cerebello-trigeminal-dermal dysplasia is an uncommon and underdiagnosed cause of SH; its phenotype was described by Gómez and López-Hernández. The characteristic clinical triad includes rhombencephalosynapsis, alopecia mainly bilateral parieto-occipital and trigeminal anesthesia. RS is an uncommon cerebellar malformation with absence of vermis and fusion of cerebellar hemispheres.

Alopecia can be bilateral parietal, occipital and/or symmetrical temporal and can be a neonatal finding or appear in the adolescence, thus the absence of alopecia during childhood does not exclude this syndrome. Trigeminal anesthesia affects the ophthalmic nerve (less sensibility in the forehead and cornea), which increases the risks of microtraumas, ulcers, corneal opacity and, eventually, blindness. Associated clinical features include a characteristic facial appearance, brachycephaly, strabismus, ataxia, developmental delay, short stature, and corneal opacities. Given the associated congenital anomalies, anesthetic care may be required for various surgical interventions.

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