Gomez-López-Hernandez syndrome: the triad of cerebello-trigemino-dermal dysplasia

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DESCRIPTION

A 14-year-old female presented with progressive diminution of vision in the right eye. She had left-sided loss of vision since 1 year of age. The patient had aggressive behaviour with irrelevant talking. She was intellectually disabled and had a history of delayed motor and language milestones. On examination, she was short-statured and had craniofacial abnormalities in the form of midface retrusion, flat nasal bridge, low-set ears, widened philtrum, thin upper lip vermillion and hypertelorism (figure 1A). The left eye was phthletic and corneal opacity was seen in the right eye (figure 1B,C). Bilateral corneal reflexes were absent. There was partial sensory loss in the distribution of ophthalmic division (V1) of bilateral trigeminal nerves. Bilateral parietotemporal alopecia (figure 1D,E) and forehead scarring (figure 1A) were present. Cerebellar signs were present in the form of difficulty in tandem gait and dysmetria.

MRI of the brain revealed a small posterior fossa. Vermis and falc cerebrelli were absent with fusion of bilateral hypoplastic cerebellar hemispheres and midline continuation of folia suggestive of rhombencephalosynapsis (RES) (figure 2A,B). There was fusion of bilateral superior cerebellar peduncles and thalami (figure 2C,D). Bilateral middle cerebellar peduncles were anteroposteriorly directed.

Bilateral trigeminal nerves were small in size (L>R) (figure 2E). The fourth ventricle was abnormal with low-lying fastigium (figure 2F). Brachyturricephalic skull with diffuse calvarial thickening was seen (figure 1F).

The patient’s clinical and imaging findings suggested a diagnosis of Gomez-Lopez-Hernandez syndrome (GLHS), also known as cerebello-trigemino-dermal dysplasia. It is a rare neurocutaneous syndrome, and approximately 73 cases have been reported to the best of our knowledge. GLHS is characterised by triad of RES, partial alopecia and trigeminal anaesthesia. However, trigeminal anaesthesia is present only in 50%-60% of cases. In the absence of trigeminal anaesthesia, the presence of other craniofacial abnormalities can also suggest the diagnosis of GLHS. Craniofacial abnormalities include brachyturricephaly, midface retrusion, hypertelorism, widened philtrum and low-set ears. Other clinical features include intellectual disability, motor delay, strabismus, ataxia, hypotonia, seizures and characteristic head shaking in ‘figure-of-eight’ pattern. Our patient did not have seizures or any stereotypical movements. Trigeminal anaesthesia...
can cause repeated unaware injuries which can lead to forehead scarring, corneal opacities and ptosis bulbi in severe cases. Aqueduct stenosis, hydrocephalus, absent septum pellucidum, absent olfactory bulbs, absent or hypoplastic trigeminal nerves, and dentate or tonsillar fusion can also be seen. Other systemic abnormalities such as absent kidney, neurogenic bladder, cryptorchidism and hypoplastic labia majora can also be associated.

The genetic basis of GLHS is still unknown. Defects in dorsoventral patterning can give rise to RES. GLHS is the most common syndromic association of RES. Insult to the embryo at 28–44 days can cause developmental arrest of part of ectoderm from which alar plate of rhombencephalon, overlying epidermis, trigeminal nucleus and trigeminal placodes arise, which can give rise to constellation of findings of GLHS. There have been reports of GLHS in children of mothers with misoprostol exposure in first trimester, substance abuse, assisted reproduction and maternal diabetes, which suggest the role of environmental and epigenetic mechanisms in its pathogenesis.

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