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Goldenhar Syndrome: Morphological appearance

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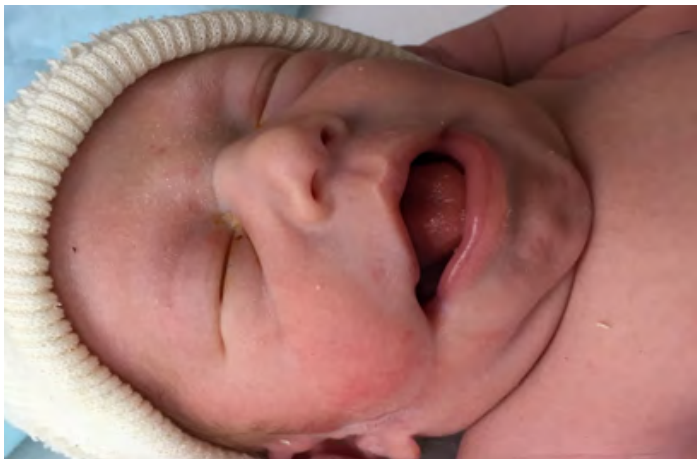
1. Clinical Image

A 14-day-old newborn, born after a monitored pregnancy carried out until 39 weeks of gestation, presents with facial dysmorphia (Figure 1) characterized by right hemifacial hypoplasia and pre-auricular diverticula. Orbital-cerebral imaging revealed left-sided facial asymmetry, hypoplasia of the malar bone, cheek soft tissues, nasal fossae, and agenesis of the corpus callosum. Additionally, general assessment did not reveal any cardiac, vertebral, or renal malformations. The diagnosis of Goldenhar Syndrome was confirmed through molecular biology.

The Goldenhar syndrome, also termed oculo-auriculo-vertebral syndrome, is a rare polymalformative disorder linked to aberrant development of the first branchial arches. Common features include periorbital dermoids or dermolipomas, preauricular skin diverticulae, ear auricle shape anomalies, conductive hypoacusis, facial structure abnormalities, and occasional vertebral anomalies. Moreover, it can co-occur with cardiac malformations and intellectual disability. This condition underscores the complexity of craniofacial development and highlights the importance of comprehensive medical assessment and management strategies for affected individuals. In addition to these anomalies, one may observe a palpebral fissure, Duane syndrome, or occasionally microphthalmia or anophthalmia [1-2].

By integrating advanced medical technologies, personalized treatment plans, and supportive care, healthcare professionals can optimize outcomes and improve the quality of life for individuals affected by Goldenhar syndrome. This underscores the importance of ongoing research and collaboration to enhance our understanding and management of this rare condition.

2. Keywords: Goldenhar Syndrome; Oculo-auriculo-vertebral syndrome



Figures 1: Morphological aspect of goldenhar syndrome.

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