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SCIENTIFIC LETTER

Goldenhar Syndrome with Imperforate Anus: New Association or Coincidence!

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To the Editor: A 12-y-old boy was brought with complaints of poor scholastic performance. He was second born to a non-consanguineous couple with an uneventful antenatal and perinatal period. He had imperforate anus at birth, which was operated on D3 of life, without any major complications in the peri-operative period. There was no history of neurological or psychiatric illness in other family members. On examination, he had facial asymmetry, microsomia, mandibular hypoplasia, epibulbar dermoid in the left eye, right pre-auricular skin tags, right pre-auricular sinus and thoracolumbar scoliosis (Supplementary Fig. 1). Systemic examination was unremarkable. Radiograph of the dorsolumbar spine showed scoliosis. Ultrasound abdomen, magnetic resonance imaging of the brain and hearing assessment was normal. He had moderate intellectual disability (intelligence quotient with Vineland social maturity scale was 47). Based of clinical examination, a diagnosis of Goldenhar syndrome was concluded.

Goldenhar syndrome is a rare congenital disorder characterized by incomplete development of first and second branchial arch derivatives. It was first described by an ophthalmologist, Maurice Goldenhar in 1952 and has many synonyms in the literature including Goldenhar-Gorlin syndrome, oculo-auriculo-vertebral syndrome and facio-auriculo-vertebral syndrome [1]. Etiology is obscure and is most often attributed to the disruption of blood flow during fetal development. Most cases are sporadic with no significant family history. Diagnosis is usually made by the OMENS (orbit, mandible, ear, nerve, and soft tissue) classification system. The signs and symptoms are unilateral in 85% of the cases with right-sided predominance [2]. Systemic involvement can vary widely and can be associated with abnormalities of cardiovascular, genitourinary and central nervous systems. Associated central nervous system malformations are diffuse cerebral hypoplasia, corpus callosum dysgenesis, encephalocele, Arnold–Chiari malformation, hydrocephalus, absence of septum pellucidum, holoprosencephaly and facial palsy that can lead to a varying degree of microcephaly, developmental delay and intellectual disability [1, 3]. Gastrointestinal malformations are seen in 11.5% cases and include esophageal and duodenal atresia, esophageal and pyloric stenosis, diaphragmatic hernia and intestinal volvulus [4, 5]. Cohen and colleagues described anorectal malformation in two children (out of 86) but they did not mention imperforate anus in their series [4]. To the best of our knowledge, Goldenhar syndrome with the imperforate anus is never reported in the literature and this can be a new association or coincidence.

Compliance with Ethical Standards

Conflict of Interest None.

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