PICTORIAL CME

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Goldenhar Syndrome: A Rare Presentation to the Physician

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Key words: External auditory canal atresia, Treacher-Collins syndrome, Pierre-Robin Syndrome, Crouzon syndrome, Goldenhar Syndrome.

An 18-year-old male delivered vaginally at term but with late cry after 5 minutes and delayed milestones, but presently fully independent in his daily activities of living, studying in high school with average academic performance, and normal social, motor, and language skills. He gave history of cleft lip operated at 1 year of age, branchial fistula operated at 3 years of age, cryptorchidism operated at 5 years of age, and left auditory canal atresia with reconstruction surgery at 12 years of age to create an external ear. He had two later siblings of which one sibling had died in infancy due to pneumonia. None of the siblings or any of his relatives had history of any similar affliction.

Clinical examination revealed facial asymmetry, low hair line, prognathism, no external auditory meatus on the right side, a reconstructed external ear on the left side, pre-auricular skin tags bilaterally, and genitalia underdeveloped for his age. X-ray of the cervical spine revealed synostosis of the upper 3 cervical vertebrae. Our patient had all the features of Goldenhar syndrome.

Few entities like Crouzon, Treacher-Collins, Pierre-Robins, and Goldenhar Syndromes have atresia of the external auditory canal as a common feature. Crouzon syndrome is inherited in an autosomal dominant pattern and is due to a

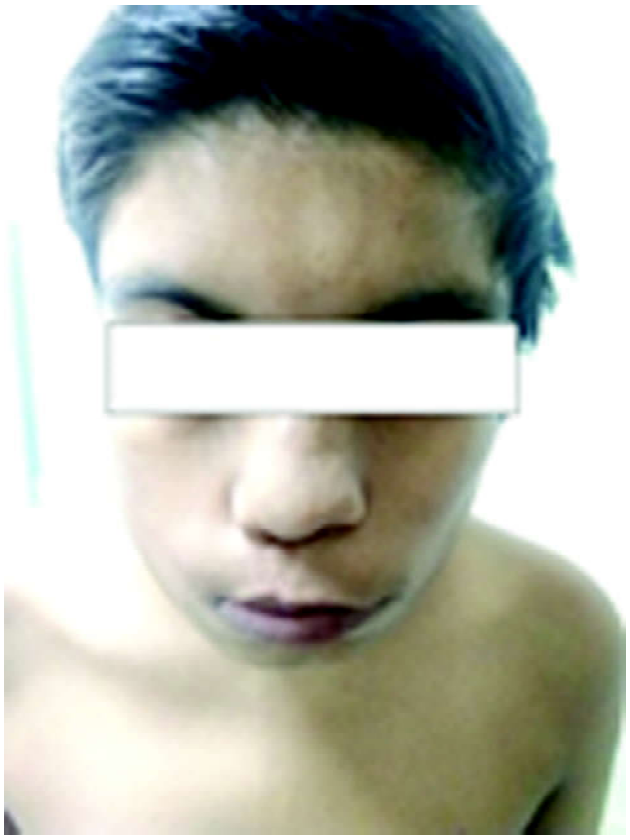

Fig. 1: *Asymmetry of the face.*

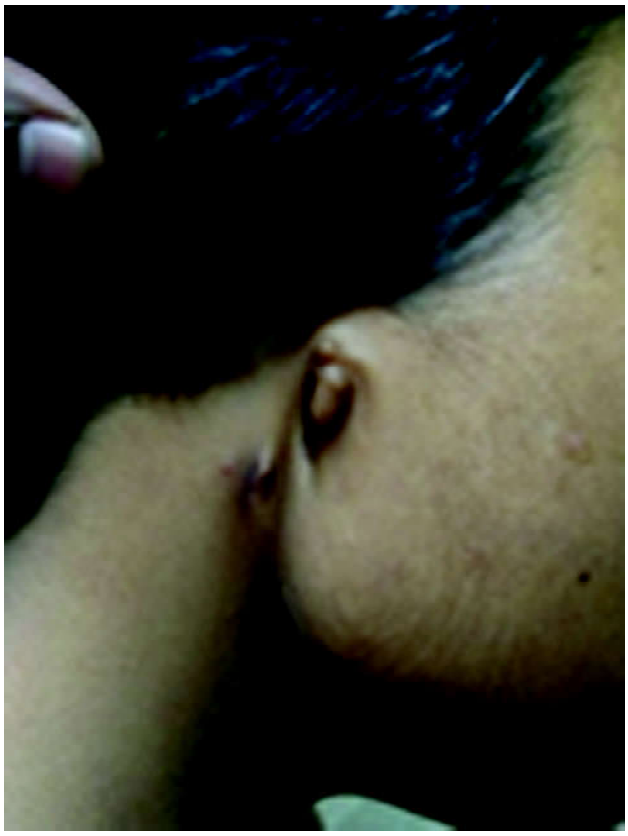

Fig. 2: *Absent external auditory meatus with skin tags.*

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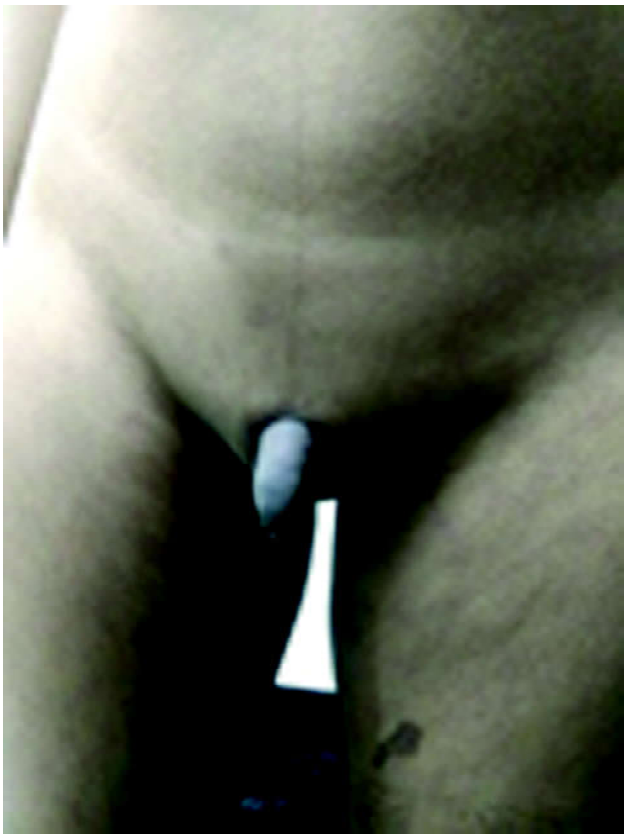

Fig. 3: *Under-developed genitals.*

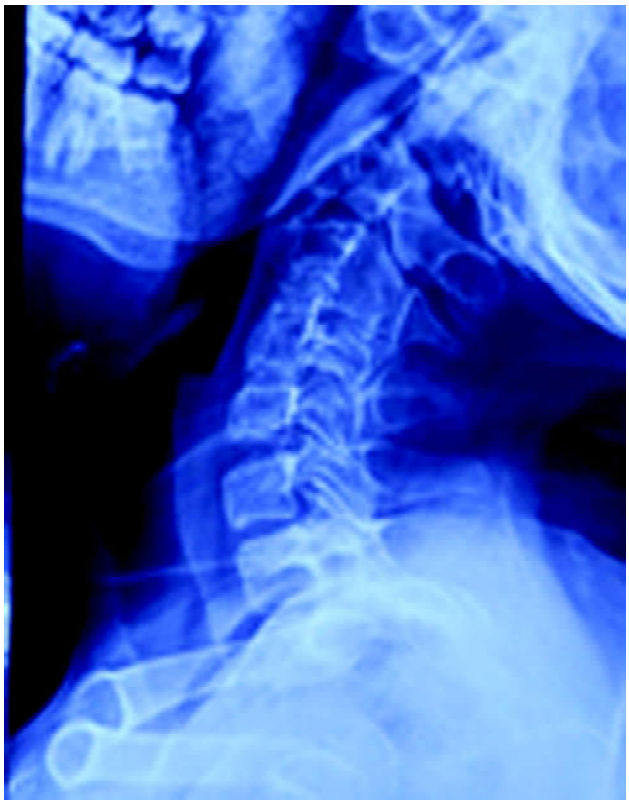

Fig. 4: *X-ray of cervical spine in Lateral view showing fusion of the 2nd, 3rd and 4th cervical vertebrae.*

mutation in fibroblast growth factor gone. It is characterised by premature craniosynostoses giving rise to cloverleaf skull. Exophthalmos, midfacial hypoplasia, Chiari I malformations (70% of cases), hydrocephalus, and stylohyoid ligament calcification (up to 50%) may also be present in patients[1].

Treacher-Collins syndrome, or mandibulofacial dysostosis, an autosomal dominant genetic abnormality, resulting from bilateral malformations of first and second branchial arches[2] due to mutations of the *TCOF1* gene, *POLR1C* and *POLR1D* [3], is characterised by dental and mandibular abnormalities including cleft palate, micrognathia, and zygomatic arch abnormalities. Patients have an obliterated nasofrontal angle. Spinal abnormalities are uncommon.

Pierre-Robin Syndrome (PRS) is the result of a sequence of events, and hence Robin Sequence should be the preferred term. The primary defect is abnormal development of the first pharyngeal arch causing a retrognathic mandible which prevents the tongue from descending which in turn prevents fusion of palatal shelves. Glossoptosis (retraction of tongue), high or U-shaped palate, micrognathia, and airway obstruction are the outcomes of this sequence. Polyhydramnios in mother is another result due to diminished swallowing in the foetus and its presence should prompt search for PRS[4].

Goldenhar syndrome, also called oculo-auriculo-vertebral spectrum (OAVS) is found in 1 in 3,000 - 5,000 newborns with a male to female ratio of 3:2. It is characterised by ear anomalies, commonest among which are pre-auricular appendages or skin tags, unilateral microphthalmia or anophthalmia, hemifacial microsomia, cleft palate, urogenital abnormalities, and asymmetry of skull and spinal anomalies[5,6]. Intelligence is usually normal. Radiological delineation of middle ear cavity volume, ossicles, inner ear structure, and course of internal carotid artery and facial nerve is important in surgical management of these cases[7].

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