CASE REPORT

Goldenhar Syndrome associated with Vestibular Fistula and Esophageal Atresia – A Rare Association

Dinesh Kumar Barolia, Sunil Kumar Mehra, Vinita Chaturvedi, Gurudatt Raipuria, Aditya Pratap Singh, Abhishek Kumar Rai
Department of Pediatric Surgery, SMS Medical College Jaipur, Rajasthan, India.


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ABSTRACT

Goldenhar syndrome is associated with various other congenital anomalies of gastro-intestinal tract, heart, kidney and genitourinary system. We report a rare association of Goldenhar syndrome with vestibular fistula and esophageal atresia.

Key words: Facial cleft, Goldenhar syndrome, esophageal atresia, Vestibular fistula.

INTRODUCTION

Goldenhar syndrome, also known as oculo-auriculo-vertebral syndrome, is a rare congenital anomaly with an incidence of 1:3500 to 1:5600 live births. [1] It is commoner among males with male female ratio reported as 3:2. We report a Goldenhar syndrome in a female child associated with vestibular fistula and esophageal atresia with tracheo-esophageal fistula which is a very rare entity.

CASE REPORT

A female neonate born prematurely at 32-week gestational age to a P9G3 mother, weighing 1700g weight was admitted to our Department. Seven of her siblings had expired previously owing to prematurity and low birth weight. The chief complaints were excessive frothy secretions from mouth, abdominal distension, respiratory distress, absent anal opening, and passing meconium from vaginal vestibule. Facial anomalies on examination included right lateral facial cleft, two pre-aureicular skin tags on the right side, one skin tag over the right cheek and two pre-aureicular skin tags on the left side [Fig.1]. Buttocks were flat and anal opening was absent; the meconium was coming out from vaginal vestibule. There was copious oral secretion, so oral suction was done. The diagnosis of esophageal atresia and tracheo-esophageal fistula was confirmed by the non-negotiation of the red rubber catheter into the stomach and gas-filled bowel on babygram (Fig.2).

Figure 1: showed right lateral facial cleft with pre-aureicular skin tag.

There were multiple cervical and vertebral anomalies and absence of multiple ribs on the right side. TORCH profiles of both the mother and the neonate were positive for cytomegalovirus (CMV) and rubella virus. She expired on second day of life before any
surgical intervention. Her parents were counseled about TORCH infection, treatment, risk, and prognosis for the future pregnancy.

Figure 2: showed cervical and thoracic vertebral anomaly, red rubber catheter could not negotiated in oesophagus.

**DISCUSSION**

Goldenhar syndrome is a birth defect resulting from the mal-development of the first two branchial arches, with incomplete development of the ear, nose, soft palate, lip and mandible [2-4]. The phenotype is highly variable. The classic triad is mandibular hypoplasia resulting in facial asymmetry, ear and eye malformation and vertebral anomalies. [5] Eye anomalies include epibulbar dermoid, coloboma, one-sided microphthalmos and lipodermoid formations. Ear anomalies are small ear, dropped ear, pre-auricular skin tags and fistulas. Along with these anomalies, Goldenhar syndrome may be associated with hemi-vertebrae, hypoplastic mandible, cleft palate, abnormal hands or fingers, pulmonary hypoplasia, Tetralogy of Fallot, ventricular septal defect, mental retardation, lymphomas in corpus callosum, agenesis of single kidney, ectopic kidney, duplication of ureter, hydrenephrosis, hydrouréter, and genitourinary system anomalies [2,3].

Ingestion of drugs such as thalidomide, retinoic acid, tamoxifen and cocaine by pregnant mothers may be related to the development of Goldenhar syndrome. [6] Maternal diabetes, rubella and influenza have also been suggested as etiological factors. [7] CMV and rubella infections may have been the etiological factor in the index case reported here. Gorlin and Pindborg suggested that during embryological development, mesoblasts become affected which leads to abnormal development of branchial and vertebral system causing Goldenhar syndrome. [3] Tracheo-esophageal fistula, anomalies of the urogenital system, ureteropelvic junction obstruction and imperforate anus with or without rectovaginal fistula also associated with this syndrome. [8]

**Consent:** Authors have submitted signed consent form from legal guardian of the patient and available with editorial office.

**Authors’ contribution:** All the authors equally contributed in concept, design, drafting of manuscript, and approved final version of the manuscript.

**REFERENCES**