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Case Report

Nasal polyposis with dextrocardia, pulmonary agenesis and microtia: Goldenhar syndrome - a case report

Preeti Dhingra*, Christopher de Souza, Kamal Parsram

Department of ENT and Head Neck Surgery, Lilavati Hospital and Research Centre, Mumbai, Maharashtra, India

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***Correspondence:**
Dr. Preeti Dhingra,
E-mail: drpreetidhingra@gmail.com

ABSTRACT

Its a case report of, Goldenhar syndrome (GS), a rare syndrome with right ear microtia, atresia of the external auditory canal, nasal polyposis, pulmonary agenesis, spine abnormalities and dextrocardia. This case, has showed the involvement of nose in the form of nasal polyposis in GS, which has never been reported in the literature so far.

Keywords: Goldenhar syndrome, Dextrocardia, Pulmonary agenesis

INTRODUCTION

Goldenhar syndrome (GS) is a rare congenital entity caused due to defective development of first and second branchial arches.[1] This syndrome is a complex disorder with variable presentation involving heart, lungs, face, ears, eyes, and vertebrae. The exact etiology of the syndrome is not known

This case report of GS is probably among the few cases reported in literature showing the association of dextrocardia with pulmonary agenesis and microtia in GS. To best of our knowledge, there has been no case report of nasal involvement in Goldenhar syndrome.

CASE REPORT

21-year-old male patient came to ENT department with history of nasal blockage right side, nasal discharge right side and snoring since one year. There was no h/o pain, nasal bleeding, chest pain, breathlessness, palpitations. There were h/o repeated episodes of cold. There was h/o decrease hearing right ear since birth with facial deformity There was no significant family history. He was born at full term after a normal delivery and both parents were nonconsanguineous.

Examination of the patient revealed prognathic features with web neck and hypoplasia of mandible. Patient had kyphoscoliosis with convexity to right side, atrophy of pectoralis major muscle on right side and marfanoid features like high arch palate. Eye examination showed hypertelorism. Examination of ears showed low set ears, pre auricular tag right side, microtia right ear, absence of external auditory canal (Figure 1). Cardiovascular examination showed hearts sounds on right side. Respiratory examination showed clear chest on left side with absence of breath sounds on right side. The patient's mental development and neurological examination was normal.

Nasal examination on anterior rhinoscopy showed polyp in the right nostril with deviated nasal septum towards left side. Oral cavity examination showed large polyp hanging in the oropharynx, pushing soft palate anteriorly and uvula to the left side. Patient had muffled voice due to mass in the oropharynx.

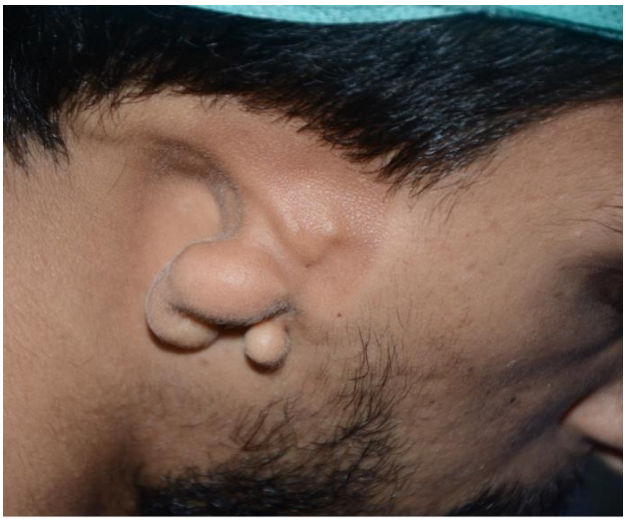

Figure 1: Clinical features of patients with low set ears, pre auricular tag, microtia, absence of external auditory canal, hypoplastic maxilla.

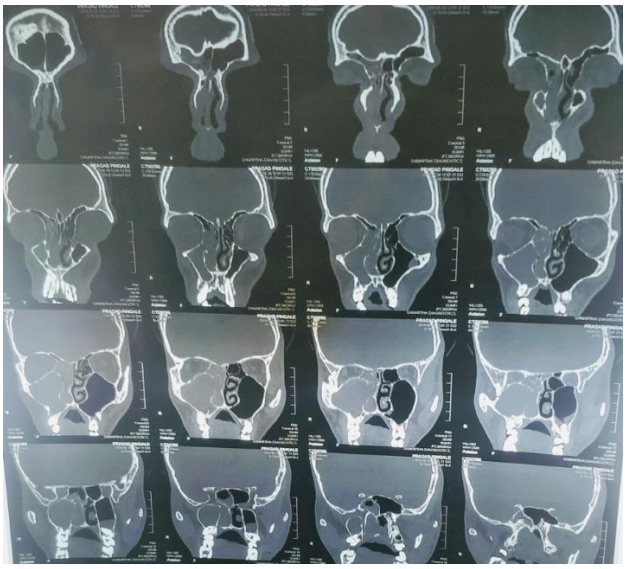

Figure 2: CT scan of nose and paranasal sinuses, showing pansinusitis right side with osteomeatal complex blockage and deviated nasal septum towards right side.

Based on the patient's clinical features, signs and other associated abnormalities, we made the provisional diagnosis of this disorder being Goldenhar syndrome. Patient was sent for CT scan nose and paranasal sinuses with showed large antrochoanal polyp in the right nostril extending into nasopharynx and oropharynx with right osteomeatal complex blockage and deviated nasal septum (Figure 2).

Patient was posted for Functional endoscopic sinus surgery under GA. All the routine general anaesthesia investigations were carried out.

Chest X-ray was done which showed agenesis of right lobe of lung (Figure 3).

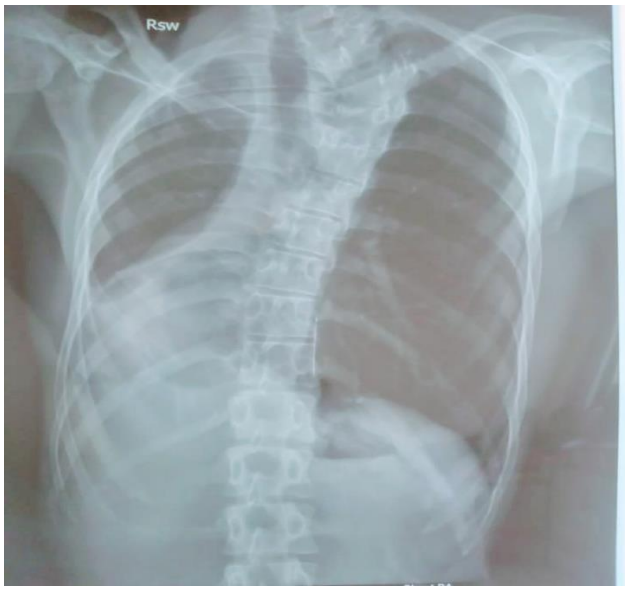

Figure 3: X ray chest showing agenesis of right lung.

Echocardiography was done which showed dextrocardia and mild prolapse of tricuspid valve LV size was normal with good LV function. Also, normal size of RA and RV.

Figure 4: Right pulmonary agenesis with compensatory hyperinflation of the left lung on HRCT.

HRCT showed features suggestive of right pulmonary agenesis with compensatory hyperinflation of the left lung (Figure 4).

MRI spine showed scoliosis of the upper dorsal spine with convexity to left, fusion of the D3 and D4 vertebrae and cleft vertebra at D2 level.

MRI brain was normal except finding of lipoma in the left tentorium cerebelli.

HRCT temporal bones showed agenesis of right pinna, external auditory canal, middle ear and middle ear ossicles (Figure 5).

Audiogram was done which showed severe mixed hearing loss right ear, with normal hearing on left side.

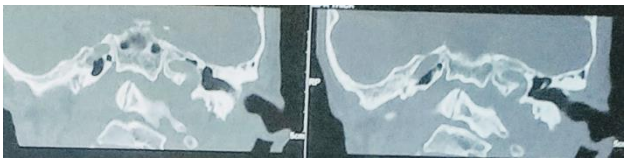

Figure 5: Agenesis of right pinna, external auditory canal, middle ear and middle ear ossicles on HRCT temporal bones.

USG abdomen was done which showed situs solitus, no situs inversus.

Spirometry and flow volume loop were done which showed severe restriction with no significant post bronchodilator reversibility. Total lung capacity was reduced (TLC), residual volume disproportionately increased to TLC with reduced diffusion capacity of lung.

Six-minute walk test showed no significant desaturation on covering 300 meters.

All these clinical features and radiological investigations confirmed the diagnosis of Goldenhar syndrome. FESS was done under GA, polyps and polypoidal mucosa were removed and tissue sent for HPE. All the sinuses were cleared. Follow up was done after 1 week, 1 month and 3 months which showed well healed nasal cavity.

DISCUSSION

Goldenhar syndrome is a rare congenital syndrome, which involves various functional and morphological abnormalities. GS is known by different names, due to the vast variations in the presentation, like first and second branchial arch syndrome, Oculoauriculovertebral dysplasia, Facioauri-culovertebral spectrum.[2]

GS was first described by ophthalmologist Maurice Goldenhar in 1952. Gorlin in 1963 coined the term oculoauriculovertebral (OAV) dysplasia. According to the literature, when malformations involves mainly the ears, jaw, mouth, and that too on one side of the body, it is called as hemifacial microsomia. If other systems are also involved like heart, vertebra, eyes, then the disorder is called GS.[3]

The prevalence rate of GS is one per 3500–7000 live births. More commonly seen in males, with male to female ratio of 3:2. The defect involves structures which arise from the first and second branchial arches, first branchial cleft, first branchial pouch, and temporal bone primordia.[4,5] Typically, the organ is either absent or underdeveloped on one side. In most cases, only one side of the face is affected, but there are some reported cases of patients has bilateral involvement also. Treacher–Collins syndrome is one of the most important differential diagnosis of GS but is usually characterised by similar features with bilateral presentation.

The exact etiology of GS is unknown, but it is considered to be multifactorial. Various theories are:

1. Sporadic: most cases of GS occurs randomly, with no cause.

2. Chromosomal anomalies: were chromosomal aberrations such as 3del (5p), del (6q), del (8q) (161), del (18q), del (22q), re and dup (22q) are known to be associated with GS.

3. Genetic: in the literature autosomal dominant, autosomal recessive inheritance patterns has been reported.

4. Antenatal use of drug like thalidomide, anticoagulants, retinoic acid, cocaine etc: during the blastogenesis phase, poor circulation of blood and focal haemorrhage in first and second branchial arches region, results in improper mesoderm formation and neural crest development.

5. Maternal diabetes, consanguineous marriage, malnutrition, exposure to tobacco during pregnancy, hypertension.

GS is characterized by wide range spectrum of signs and symptoms that varies from individual to individual. This syndrome presents with classical triad of ocular, auricular and vertebral anomalies, along with cardiac, renal and central nervous system anomalies also.[6,7]

Ear abnormalities in GS includes low set ears, microtia, atresia of the external auditory canal, agenesis of middle ear and internal ear anomalies, preauricular appendages. This patient had all these ear features of GS with profound hearing loss right ear.

Among the ocular seen anomalies seen in GS, important are epibulbar dermoids, cataract, astigmatism, microphthalmia, antimongoloid obliquity of palpebral fissures, blepharophimosis and colobomata of the upper eyelid, iris, and retina. Our patient presented only with hypertelorism.

Vertebral anomalies reported in the literature include hypoplasia of vertebra, fused vertebrae, hemivertebrae, and scoliosis or absence of certain vertebra. This patient had scoliosis of the upper dorsal spine with convexity to left, fused D3 and D4 vertebrae and cleft vertebra at D2 level.

The frequency of cardiovascular anomalies seen in GS ranges from 5% to 58%. The incidence of dextrocardia associated to situs inversus in the general population is usually 1:10,000, whereas dextrocardia with situs solitus is 1:30,000 live births and only 1:900,000 in the adult

population. Some cardiac and extracardiac abnormalities which are commonly seen in patients with Dextrocardia and situs solitus, are single ventricle, atrial septal defect, ventricular septal defect, transposition of the great arteries, pulmonary hypoplasia, tracheoesophageal fistula, imperforate anus, spina bifida and Kartagener syndrome. Usually dextrocardia with situs inversus is not associated with congenital heart disease. Some patients of dextrocardia with situs inversus has Kartagener's syndrome, characterized by chronic sinusitis, nasal polyposis and bronchiectasis. In our case, except dextrocardia and mild prolapse of tricuspid valve, no other cardiac anomaly was found.

Agenesis or hypoplasia of the lungs is a part of GS. The term expanded Goldenhar complex is used to for GS with pulmonary agenesis or hypoplasia. Usually these patients have repeated episodes of respiratory distress in the neonatal period. Our patient had right pulmonary agenesis seen on chest X-ray and HRCT, but is diagnosed accidentally when he got admitted in hospital for nasal surgery. This patient had never complained of any respiratory issues till age. Urological and gastrointestinal anomilies which are reported in GS are ectopic kidneys, uretropelvic junction obstruction, and imperforate anus. In this patient, no gastric or intestinal anomaly was seen.

As far as face is concerned, asymmetry of face, hypoplastic mandible and maxilla are characteristic features of GS.[8,9] Cleft lip is observed in 5% of the cases. Other facial abnormalities in GS include, zygomatic bone hypoplasia, high arched palate, micrognathia, dental malalignment, and underdeveloped facial muscles. Our case classically had facial deviation to right side with mandible hypoplasia, high arched palate, malalignment of jaw, overcrowding of upper teeth.

According to the recent literature, the involvement of nose has not been seen in this syndrome. This patient had pansinusitis and large antrochoanal polyp right nostril which was extending into nasopharynx and oropharynx. Also, there was septal deviation towards right side along with spur. The reason of antrochoanal polyp and blocked osteomeatal complex in this patient could be due repeated episodes of cold and decrease mucociliary clearance.

The diagnosis of this condition is not always easy due to its varied clinical presentations. And also, there is no specific diagnostic test for GS. Diagnosis is usually by thorough history and physical examination of the patient. Early diagnosis of this condition is important for the proper management of the patients and genetic counseling. As GS affects several systems of the body, the management of GS patients is complex and requires multiple specialities for the proper development and rehabilitation of the patient.[10] Also it requires long-term commitment from the patient and family. The most important among all is the emotional and psychological support to the patient. Depending on the disease, some cases require surgery and some just need careful observation. For example, in case of patients with mandibular and maxillary hypoplasia, reconstruction can be done. For microtia pinna reconstruction can be performed.

CONCLUSION

GS is a rare congenital syndrome characterised by craniofacial anomalies. The involvement of nose in GS has never been reported in the literature, so our case is the first one to report the involvement of nose in GS. In this case it's important to differentiate GS from Kartagner syndrome.

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