Goldenhar Syndrome: Case Report

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Abstract

Goldenhar syndrome or fascio-auriculo-vertebral dysplasia or oculo-auriculo-vertebral syndrome is a sporadic or autosomal dominant inherited genetic rare syndrome characterized by mandibular hypoplasia, facial asymmetry, low set ear or atresia of ear canal, preauricular skin tags, hemi vertebra in cervical region, epibulbar dermoid, coloboma of upper eyelid, limb dermoids, cardiac abnormalities and other systemic abnormalities includes facial involvement, predisposing to the right side or there may be a more complex phenotypic abnormality with the skeletal, cardiac, renal and pulmonary systems. Central nervous system involvement are common with these patients, particularly there are higher chances with ophthalmologic anomalies. 50% of the patients with Goldenhar would have either conductive and/or sensorineural hearing loss. This case report describes a typical 40-year-old female patient who presented to the hospital with auricular abnormality and diminished hearing and was found to have the fascioauriculo-vertebral dysplasia spectrum of this syndrome and hypothyroidism. Diagnosis was based principally on clinical aspects. Radiology, laboratory findings, otorhinolaryngologic evaluations were important in reaching a definitive diagnosis.

Management depends on the patient’s age and systemic clinical manifestations, with a multidisciplinary approach often being required.

Keywords: Autosomal dominant, Goldenhar syndrome, Hypothyroidism, Isolated, Preauricular tag

INTRODUCTION

The incidence of Goldenhar syndrome (GHS) ranges from 1 in 5,600 to 1 in >20,000 live births. It is more common among males, with male:female ratio of 3:2.1 It involves abnormalities in the first and second branchial arches. The etiology of the syndrome is heterogeneous.

The syndrome may range from mild to severe forms; two pathophysiologic mechanisms have been proposed for the oculo-auriculo-vertebral syndrome (OAVS) which includes, reduction in the blood flow resulting in focal hemorrhage in the first and second branchial arches around 30-45 days of gestational age, which lies in the blastogenesis period2 which are responsible for external ear abnormalities, as development of anterior ear primordium takes place from first branchial arch, and the second arch modifies to posterior ear primordium. Furthermore, the outer ear canal is derived from the dorsal portion of first branchial cleft.3,4 The etiology may be related to an anomaly in migration defect of neural crest cells or could be due to predisposed genetic determinant. Some studies document families with autosomal recessive or dominant inheritance, it contains several descriptions of chromosomal anomalies and gestational exposure that mimic its phenotype.5 Facial asymmetry is reported in about 70% of the cases, whereas ear anomalies in 83% and eye malformations in 66%. Ear abnormalities, ranged from anotia to preauricular tag.6 Ear anomalies are frequent and varied and are clinically heterogeneous, and there are no clear guidelines available in the literature on the diagnostic criteria. Often there is no correlation among the findings from external, middle, and inner ear.

CASE REPORT

A 40-year-old female patient resident of Jalgaon came to Surgery Department with complaints of decreased hearing and tinnitus since 1 year that was gradually increasing. There was a history of congenital right pinna atresia, facial asymmetry on the right side with deviation of the angle of mouth. On examination, patient had preauricular tags on the right side with left ear anatomically normal.
There was no mental retardation. No evidence of sinuses was seen. Her audiometry shows bilateral profound mixed hearing loss. Patient had been treated for atrial fibrillation in the past. 2D ECHO was suggestive of 70% ejection fraction and rest findings were normal. Patient had scoliosis and hemi vertebra in the cervical region patient was on thyroxine tablets, and she was not diagnosed having diabetes mellitus, epilepsy, tuberculosis or trauma in the past.

**DISCUSSION**

GHS, fascio-auriculo-vertebral dysplasia or OAVS is a sporadic or autosomal dominant inherited genetic rare syndrome characterized by mandibular hypoplasia, facial asymmetry, low set ear or atresia of ear canal, preauricular skin tags, hemi vertebra in cervical region, epibulbar dermoid, coloboma of upper “eye lid,” limb dermoids, cardiac abnormalities. And other systemic abnormalities facial involvement is usually asymmetric, occurring mostly on the right side or it might be associated with a more complex phenotype resulting in gross manifestations in skeletal, cardiac, renal and pulmonary systems.5,7

Differential diagnosis with the similar features include, syndromes that are derived from aberrations in the first and second branchial arches during embynic development, are included in the spectrum of GHS, which also includes Treacher-Collins syndrome (TCS). Differentiating features amongst GHS and TCS include facial asymmetry and hypoplasia of the malar bones.8 The TCS presents with downward slanting palpebral fissures, and hypoplastic changes in colobomas, zygomatic and mandibular zones, along with reduced cilia over lower eyelid cilia, and congenital abnormalities of ears.9

The evaluation protocol of patients include identification; prenatal, perinatal, postnatal and complete family history, followed by complete general examination and systemic examination of the external ear and facial features. Complimentary evaluation included: Hematological, hormonal study depending on presentation and chromosome studies. Audiological evaluation includes pure tone audiometry, speech audiometry, tympanometry, stapedius reflex measurement, and brainstem evoked response. 2D ECHO for cardiac abnormalities, skull, facial, and upper limbs spine X-rays; temporal bone computed tomography (CT) scan, CT spine, magnetic resonance imaging if required. Patient is followed up.

Treatment depends on the patient’s age and systemic manifestations, but generally requires a multidisciplinary approach.1,10 Varies according to age of the patient: Between 2 and 4 years, no treatment is necessary for patients who are mildly affected. Reconstruction by rib bone graft and lengthening of underdeveloped mandible by a bone distraction device is recommended in severe underdeveloped mandible, modifications in the growth of teeth are done with the assistance from orthodontics. Amongst, 6-8 years reconstruction of the external ear is done in several stages over the period of 6-12 months.

In 8-10 years, asymmetry of cheek is to be reconstructed. This may be the most important stage in entire treatment program, in terms of physical appearance. In mild involvement no surgery is required while jaw surgery may be done in teenagers. Advantage is that the patient is fully grown. Surgical correction includes temporary wiring of the jaws.

My patient presented with hypothyroidism and cardiac abnormality along with typical features of fascio-auricular-vertebral dysplasia spectrum of GHS. Despite so many features, association of various endocrinological abnormalities (hypothyroidism) should be studied to improve the quality of life.

**CONCLUSION**

Endocrinological investigation is a must inpatient of GHS for better quality of life and better prognosis.

**REFERENCES**


How to cite this article: Patil NA, Patil AB. Goldenhar syndrome: Case report. IJSS Journal of Surgery. 2015;1:18-20.

Source of Support: Nil, Conflict of Interest: None declared.