Case Report

Goldenhar syndrome: case reports with review of literature

Muhammad Arif Maan, Ghufran Saeed*, Shahid Javaid Akhtar, Javed Iqbal

Department of Dermatology, Punjab Medical College Faisalabad
* Department of Dermatology, Independent Medical College Faisalabad

Abstract

Goldenhar syndrome is a rare developmental disorder affecting the first and second branchial arches. It is characterized by many malformations affecting face. We describe two siblings in a family with this rare disorder.

Key words

Goldenhar syndrome.

Introduction

Goldenhar syndrome (GS) is also known as hemifacial microsomia, oculo-auriculo-vertebral (OAV) syndrome, facioauriculovertebral syndrome, Goldenhar-Gorlin syndrome, first and second branchial arch syndrome, and oculoauriculo-vertebral dysplasia. GS was named in 1952 when Goldenhar described a number of facial problems that tend to occur together. These include:

- Opening of mouth larger and extended towards the ear on one side
- Underdevelopment of the muscles of the face, cheek bones and skin
- Small or misshapen ears
- Skin tags or pits usually in front of the ear in line with the mouth opening
- Mouth problems such as lack of saliva, problems in tongue shape or use
- Hemispinal vertebrae which are small or not completely formed on one side. Other problems that may occur in some but not all cases are eye defects, deafness, cleft palate, heart, limb or kidney.

This syndrome comprises of malformations arising from defects in the 1st and 2nd branchial arches. It is relatively common with an incidence of 1:5600. However, the exact genetic defect is not known.

We report here two cases who presented at Department of Dermatology, DHQ Hospital Punjab Medical College, Faisalabad.

Case 1

An 11-year-old boy presented with microsomia on left side. There was an epibulbar dermoid cyst in right eye in lateral canthus. There were pits and accessory tragi on right side. Teeth were conical and widely spaced. There was no family history of similar defects.

On examination there was no systemic abnormality except slight slurring of speech.
A 2-year-old girl presented with microsomia and accessory tragi on right side and epibulbar dermoid cyst on left side. Mother gave history of difficulty in feeding. There was no family history of birth defects. Systemic examination revealed no abnormalities.

**Discussion**

Goldenhar syndrome (GS) is a well-recognised developmental disorder involving first and second branchial arches and is characterized by considerable phenotypic variability.¹⁻¹²

In 1995, Gulf War veterans showed concerns about the unusual number of their children with Goldenhar syndrome 14.7/100,000 as compared to infants of veterans who did not deploy to Persian gulf 4.8/100,000.¹³

**Etiology and embryopathogenesis**

Majority of cases are sporadic and there is a very small chance of familial occurrence. In many reports autosomal recessive or dominant inheritance has been suggested.¹⁴ Multifactorial inheritance (interaction of many genes, possibly in combination with environmental factors such as chemical exposure). Early developmental arrest in ⁴ᵗʰ week probably due to decrease in blood flow, drugs such as primidone and vitamin-A and diseases such as diabetes, haemorrhagic incidence during ⁸ᵗʰ to ¹²ᵗʰ week involving ¹ˢᵗ and ²ⁿᵈ branchial arches, chromosomal non-dysjunction and subsequent mesodermal tethering might be the underlying mechanisms.

**Laboratory investigations**

Currently no genetic/DNA test is available; hence prenatal diagnosis is not possible. Good quality ultrasound may detect the obvious defect. High resolution computed
tomography especially for inner ear, middle ear and vertebral defects and magnetic resonance imaging might be helpful in the diagnosis.

**Treatment**

Children with Goldenhar syndrome often need many surgeries including plastic surgery to fix the jaw, cheeks and ears. Specialized dental care, hearing aids, speech therapy, physiotherapy may be required.

**Prognosis**

Low IQ, systemic involvement may show guarded prognosis. Otherwise a normal life span is expected.

**References**