Goldenhar's Syndrome

Synonyms: hemifacial microsomia, oculo-auriculo-vertebral dysplasia, oculo-auriculo-vertebral spectrum

Goldenhar's syndrome is a birth defect resulting from the maldevelopment of the first two branchial arches, with incomplete development of the ear, nose, soft palate, lip and mandible. The phenotype is highly variable.

The classic triad is mandibular hypoplasia resulting in facial asymmetry, ear and/or eye malformation and vertebral anomalies. Goldenhar's syndrome is often characterised by a combination of anomalies including dermal epibulbar cysts, auricular appendices and malformations of the ears.

Epidemiology

- Incidence is approximately 1 in 35,000 to 1 in 56,000 births. However, the incidence is higher (around 1 in 1,000) in those children with congenital deafness.\(^1\)
- The male:female ratio is 3:2.
- Most cases are sporadic. However, there are rare familial cases that exhibit autosomal dominant, autosomal recessive and multifactorial modes of inheritance.

Aetiology

- Ingestion of drugs such as thalidomide, retinoic acid, tamoxifen and cocaine by pregnant mothers may be related to the development of Goldenhar's syndrome.\(^2\)
- Maternal diabetes, rubella and influenza have also been suggested as aetiological factors.\(^2\)
- Heavy alcohol consumption during pregnancy is also one of the aetiologies of this syndrome.\(^3\)

Presentation

The degree of abnormalities between cases varies from severe to mild.\(^4\) The main features of this condition are unilateral underdevelopment of one ear (which may even not be present) associated with underdevelopment of the jaw and cheek on the same side of the face. The following may also occur:

- Facial hypoplasia and asymmetry: one side small (hemifacial microsomia), with hypoplastic mandible, malar, maxilla and facial muscles.
- Low-set, usually malformed ear: unilateral deformity of the external ear, which may include preauricular cartilage tags, external meatal abnormalities, conductive deafness or even absence of the ear (anotia).
- Sensorineural deafness.
- Mouth: macrostomia, cleft lip and/or cleft palate (10%), soft palate malfunction, agenesis of the parotid, tracheo-oesophageal fistula.
- Coloboma: coloboma of the eyelids/iris/retina/choroid - most commonly of the upper eyelid, strabismus, epibulbar dermoid cyst, microphthalmia, anophthalmia, blepharoptosis.
- Vertebral abnormalities: hemivertebra or block vertebra, vertebral hypoplasia, absent ribs, short neck.
- Midline facial cleft.
- Cardiac defects: tetralogy of Fallot and ventricular septal defects are the most common cardiovascular anomalies. Patent ductus arteriosus, coarctation of the aorta and pulmonary stenosis can also occur.\(^5\)
- Respiratory: lung hypoplasia, increased risk of developing obstructive sleep apnoea.\(^6\)
- Urogenital: ectopic kidney, renal agenesis, hydronephrosis, multicystic dysplastic kidney, malformed or absent uterus.
- Central nervous system defects: learning difficulties (these may be as a result of deafness but there may be other structural problems such as hydrocephalus, Arnold-Chiari malformation, occipital encephalocele, agenesis of corpus callosum, hypoplasia of septum pellucidum).\(^7\)
- There may also be facial nerve palsy.

Investigations

There is no single test to diagnose Goldenhar's syndrome. The diagnosis is a clinical one.

- Once a child is diagnosed with this syndrome, additional tests should be performed which usually include:\(^8\)
  - Hearing test to determine if there is hearing loss.
  - X-ray of the spine to determine if there are vertebral problems, including scoliosis.
  - Renal ultrasound and echocardiogram are usually recommended, due to the increased risk for birth defects in these areas.
  - Eye assessment by an ophthalmologist.

Management
In uncomplicated cases, the treatment of the syndrome varies with age and systemic associations and it is mainly cosmetic.

- Surgical management involves craniofacial reconstruction.
- Reconstructive surgery of the external ear may be undertaken, usually around 6-8 years of age.
- Jaw reconstructive surgery may be needed for some patients.
- Supportive multidisciplinary management should be arranged, including departments such as paediatric cardiology, audiology, ophthalmology and plastic surgery.

**Prognosis**

Prognosis is variable and depends on the presence and severity of associated cardiovascular, neurological and other complications.

**Further reading & references**

- Goldenhar Family Support Group (UK)


**Disclaimer:** This article is for information only and should not be used for the diagnosis or treatment of medical conditions. Patient Platform Limited has used all reasonable care in compiling the information but makes no warranty as to its accuracy. Consult a doctor or other healthcare professional for diagnosis and treatment of medical conditions. For details see our [conditions](https://www.patient.info/conditions).

**Author:**
Dr Louise Newson

**Peer Reviewer:**
Dr Adrian Bonsall

**Document ID:**
2205 (v23)

**Last Checked:**
23/06/2015

**Next Review:**
21/06/2020

**View this article online at:**
[patient.info/doctor/goldenhars-syndrome](http://patient.info/doctor/goldenhars-syndrome)

Discuss Goldenhar's Syndrome and find more trusted resources at [Patient](http://patient.info/doctor/goldenhars-syndrome).