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Short Communication

Goldenhar Syndrome

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Introduction

Goldenhar syndrome is a congenital birth defect resulting from the mal-development of the first two brachial arches, which leads to incomplete development of the ear, nose, soft palate, lip and mandible[1]. The classic triad includes facial asymmetry, ear or eye malformations, and vertebral column anomalies[2]. It is also called as Occular-auriculo-vertebral Syndrome, hemifacial microsomia, oculo auriculo vertebral dysplasia, oculoauriculo vertebral spectrum etc[3]. The name came after the Ophthalmologist, Maurice Goldenhar who studied it first in 1952

Incidence

It accounts for 1 in 3000-25000 live births. Family history is rare.

Etiology

The cause of Goldenhar syndrome is idiopathic. In rare cases, about one or two percent, the condition can be inherited as a genetic disorder. In this case it is either autosomal dominant or recessive. This means that the gene or genes causing it are found on a non-sex chromosome. The genes can either be dominant or recessive, but dominant is more common.

Clinical features

The symptoms of Goldenhar and their severity are extremely variable from one individual with the syndrome to another. It usually affects one side of the face only. The most recognizable symptom is the presence of facial abnormalities, also called hemifacial microsomia. Those with Goldenhar may also have a cleft lip or cleft palate. Common signs include limbal dermoids, preauricular skin tags and strabismus[4].

Other characteristics of the syndrome include defects in the eyes, ears, and spine. This can mean cysts on the eyes, small eyes, crossed eyes, missing eyelids, small ears, missing ears, ear tags, or even hearing loss. In the spine, Goldenhar can cause incomplete development of vertebrae, or fused or missing vertebrae. Many people with the syndrome end up with scoliosis. Between five and 15 percent of people with Goldenhar have some degree of mental retardation. Some also have abnormalities in internal organs, which most commonly include congenital heart defects. Defects affecting the kidneys and limbs are rare.

- Weakness in moving the side of the face that is smaller
- The chin may be closer to the affected ear
- One corner of the mouth may be higher than the other
- Dental problems - the soft palate may move to the unaffected side of the face
- The tongue may be smaller on the affected side of the face[5]

Diagnosis

There is no genetic or chromosomal test to identify Goldenhar Syndrome. A specialist makes a diagnosis by examining an infant or child and identifying the symptoms of the syndrome.

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When it is diagnosed, the child usually needs to have further tests, such as hearing and vision tests. X-ray of the spine to check for problems with vertebrae. To look for heart or kidney problems ultrasound imaging tests of those organs can be done.

Treatment

Treatment for Goldenhar syndrome varies greatly depending on the needs of individuals. In mild cases, no treatment may be needed. If the child is having hearing impairment, they need to work with a hearing specialist or speech therapist, or may need a hearing aid. If there are vision problems, corrective surgery or glasses may be needed. Surgery may also be needed to correct heart or spinal defects. Children with mental retardation may need to work with education specialists.

Cosmetic surgeries can be performed to give a more normal appearance or to correct a cleft lip and palate. Depending on the severity of Goldenhar Syndrome, the following surgeries can be performed to correct the deformities:

- Lowering of the jaw on the affected side
- Lengthening of the lower jaw
- 3 to 4 operations to rebuild the outer ear
- Addition of bone to build up the cheeks
- Soft tissue may need to be added to the face[5]

Prognosis

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

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