Fact Sheet

Spotlight on Goldenhar Syndrome

The primary content of this article was written by Holly Ann Ismael at Health Line - http://www.healthline.com/galecontent/goldenhar-syndrome

What is Goldenhar syndrome? Dr. Maurice Goldenhar, a French ophthalmologist, first described the condition in 1952. Goldenhar syndrome is a rare congenital (present at birth) disorder that is characterized by incomplete development of the ear, eye, nose, soft palate, lip, and jaw. These physical differences have a range of severity.

Another name for Goldenhar syndrome is oculoauriculo-vertebral spectrum. This name describes the common structural problems seen with the eyes, ears, and vertebrae. The term oculo represents the eye, auriculo represents the ear, and vertebrae stands for the physical problems present in the vertebrae or the bones of the spinal column.

Goldenhar syndrome occurs in one of every 3,000 to 5,000 live births. Males are affected more frequently than females. This syndrome is seen in all ethnic groups and cultures.

What are the Features of Goldenhar Syndrome? The physical differences are typically limited to the head and bones of the spinal column and may be severe or mild. In some cases, the changes are seen on both sides of the face (bilateral). In other cases, the changes are limited to one side of the face (unilateral). It is more likely that the right side of the individual’s face will be more affected.

Facial Features: The facial bones, including the jaw bones (mandible) and cheek bones (maxilla), can be underdeveloped (hypoplasia). This underdevelopment can be limited to one side of the face. This is called hemifacial microsomia. Hemifacial microsomia can occur alone or with Goldenhar syndrome. If an individual has hemifacial microsomia without additional physical problems, the diagnosis of Goldenhar syndrome is unlikely.

Cleft lip and cleft palate can also be associated with Goldenhar syndrome. Cleft lip is an abnormal split or opening in the lip that can extend towards the nose or towards the cheek.
Cleft palate is an opening in the roof of the mouth. Individuals with Goldenhar can also have wide mouth (macrostomia).

**Vertebral Features:** The vertebral problems may result from incomplete development of the bones in the spinal column. Vertebrae can be incompletely developed (hemivertebrae), absent, or fused. Ribs can also be abnormal. Approximately 50% of individuals with Goldenhar syndrome will have curvature of the spine (scoliosis).

**Visual / Ocular Features:** Birth defects of the eye are common in Goldenhar syndrome. Cysts on the eyeball (epibulbar dermoids) occur frequently, as does microphthalmia (small eyes). Some individuals with Goldenhar syndrome have tissue missing (coloboma) from the eyelid(s). Strabismus (crossing of the eyes) is also prevalent. The child may be light sensitive (photophobia) and have decreased visual acuity that may improve with corrective lenses.

**Hearing / Otocoustic Features:** Hearing loss is common in individuals with Goldenhar syndrome due to abnormal development of the ears. The ears may be smaller than normal (microtia), or absent (anotia). Ear tags (excess pieces of skin) may be seen on the cheek next to the ear and may extend to the corner of the mouth. The shape of the ears may also be unusual. Another common problem is malformed middle ear bones (ossicles), which results in a conductive hearing loss because sound cannot be efficiently transmitted from the eardrum to the inner ear. However, malformed ossicles can be repaired or replaced at the time the atresia repair surgery is performed.

In addition there may be a missing or undersized ear canal (congenital aural atresia). If the ear canal is missing, the result is a significant conductive hearing loss, because the bone blocks the passage of sound to the eardrum and inner ear. In some cases, the ear canal is unusually narrow rather than completely closed. This is called canal stenosis. Canal stenosis also causes hearing loss and results in a higher propensity for developing ear infections (otitis media). In addition, if dead skin cells and earwax cannot drain out of the middle ear in the normal way, this can also contribute to ear infections and occasionally the formation of benign cysts (cholesteatomas).

**Cognitive and Health Features:** Other differences outside of the face and vertebra can occasionally be seen in Goldenhar syndrome. Approximately 15% of individuals with Goldenhar syndrome have an intellectual disability. The likelihood for an intellectual disability increases if the individual has microphthalmia. Heart defects and kidney defects can also occur with Goldenhar syndrome.

**What Causes Goldenhar Syndrome?** The exact cause of Goldenhar syndrome is unknown. There are most likely many factors that lead to the abnormal development of the facial tissues. In some cases the factors may be environmental in nature. For example, there are certain medications a woman can take while pregnant that can cause the baby to have the symptoms of Goldenhar syndrome. However, in the vast majority of cases, Goldenhar syndrome is not caused by something taken during pregnancy.

In other cases, normal development of the facial tissues may be disrupted by genetic factors. The exact genetic factors are unknown. Unlike some other syndromes, there has not been a
Gene identified that, if changed, causes Goldenhar syndrome. A few families in which Goldenhar syndrome occurs show an autosomal recessive inheritance pattern, while other families clearly support an autosomal dominant pattern of inheritance. However, most cases of Goldenhar syndrome are not inherited, meaning that it does not normally run in families.

Goldenhar syndrome typically occurs randomly. Doctors are often unable to explain why it occurs. Since it is sporadic in nature, if a child is diagnosed with Goldenhar syndrome, the risk for the parents to have another child with this same syndrome is low. In rare cases, one parent may have some of the physical symptoms of Goldenhar syndrome. If this is the case, the risk to have a child with the disorder may be much higher.

**Diagnosis** There is not a genetic test that can diagnose Goldenhar syndrome. The diagnosis is made by a physician when an individual has the common symptoms associated with the condition.

**Treatment and management:** Once a child is diagnosed with Goldenhar syndrome, care should be taken to complete necessary medical tests to determine the extent of the child’s physical challenges.

A hearing evaluation by an audiologist is necessary to determine if there is hearing loss. Hearing aids may be beneficial to the child. Any malformation of the outer ear and ear canal may restrict the types of hearing aids that will work for the child. Bone conduction hearing aids may be more beneficial to the child. If hearing loss is evident, the child should be referred to a certified teacher of students who are deaf/hard of hearing. Speech therapy may also be recommended.

An ophthalmologist should be consulted for a complete eye examination and needed eye care follow-up. If the child has an uncorrectable visual loss from diagnosed microphthalmia, coloboma(s) and/or other visual condition, he or she should be referred for services from a certified teacher of students with visual impairments (TVI) and a certified orientation and mobility specialist (COMS) in local early intervention and school-age programs.

X rays of the spine are recommended to determine if there are vertebral problems, and the severity. Individuals with Goldenhar syndrome should also be regularly evaluated for scoliosis. Renal ultrasounds and ultrasounds of the heart may also be recommended, due to the increased risk for birth defects in these areas. A doctor would make this recommendation. The child may also benefit from occupational and/or physical therapy.

Surgery may be required to correct the birth defects seen in Goldenhar syndrome. Surgery to correct the facial birth defects can improve appearance and function.
Resources


WEBSITES


For more information about the CO Services for Children and Youth with Combined Vision and Hearing Loss Project contact:

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Fact Sheets from the Colorado Services to Children and Youth with Combined Vision and Hearing Loss Project are to be used by both families and professionals serving individuals with vision and hearing loss. The information applies to children, birth through 21 years of age. The purpose of the Fact Sheet is to give general information on a specific topic. More specific information for an individual student can be provided through personalized technical assistance available from the project. For more information call (303) 866-6681 or (303) 866-6605. Updated: 9/11