GOLDENHAR SYNDROME

Otherwise known as?

Oculoauriculovertebral spectrum or OAVS

Signs & Symptoms

Goldenhar syndrome is a rare congenital condition that affects the development of the eyes, ears, and spine. It is considered a craniofacial syndrome, as it often involves differences in the facial structure, particularly on one side of the face. The syndrome can vary widely in its severity and presentation, even among individuals with the same condition.

Key characteristics of Goldenhar Syndrome:

1. Facial differences:

- Hemifacial microsomia: Underdevelopment of one side of the face, particularly the lower jaw and cheekbone.
- Ear anomalies: These can range from missing or underdeveloped ears to a condition called microtia (small, malformed ears) or preauricular tags (small skin tags in front of the ears).
- Eye anomalies: These may include epibulbar dermoids (benign cysts or growths on the surface of the eye), colobomas (a gap or hole in parts of the eye), or other vision problems.

2. Spinal differences:

- Vertebral anomalies such as scoliosis (curved spine) or fused vertebrae.
- Some individuals may have rib anomalies as well.

3. Other Possible Features:

- Cleft lip and/or palate: Some individuals with Goldenhar syndrome may have a cleft in the lip or palate.
- Cardiac and renal issues: Some children with the condition might experience heart or kidney difficulties, though these are less common.
- Hearing loss: Due to differences in ear formation, some individuals may experience hearing impairment, which can range from mild to severe.

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Goldenhar syndrome usually affects one side of the face more than the other, making it an asymmetrical condition. However, in about 10-33% of cases, it can be bilateral, affecting both sides of the face.

Causes (*the why*)

The exact cause of Goldenhar syndrome is not always clear, but it is believed to arise from a combination of genetic and environmental factors.

1. Genetic Factors

Goldenhar syndrome is typically thought to result from genetic mutations that affect the normal development of facial structures, especially during early foetal development. The exact genes responsible for the condition are not always identified, but some potential contributing factors include:

- Chromosomal differences: In some cases, Goldenhar syndrome may be associated with chromosomal deletions or duplications. However, chromosomal changes are not seen in all cases of the syndrome.
- Inherited mutations: The condition can sometimes be inherited in an autosomal dominant pattern, meaning a child can inherit the syndrome from an affected parent. However, most cases are sporadic (occurring by chance without a family history), and in these cases, the mutation arises as a new, random event during early development.

2. Disruption of Embryonic Development

Goldenhar syndrome is thought to result from differences in the development of the first and second branchial arches (the structures that form the face, ears, and jaw during early foetal development). The condition likely arises when these arches fail to develop normally, leading to the characteristic facial and ear differences.

Key events during early embryonic development that may contribute include:

- Incomplete fusion of the branchial arches: When the usual fusion of the structures that form the face and jaw doesn't occur, this may lead to hemifacial microsomia, where one side of the face is underdeveloped.
- Interruption of blood flow to developing tissues: Reduced blood supply to the facial tissues during early development could lead to the underdevelopment or absence of certain facial features, such as the ear or eye.

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3. Environmental Factors

Certain environmental factors may contribute to the development of Goldenhar syndrome, especially during pregnancy. These factors may disrupt the normal development of the first and second branchial arches.

4. Other Factors

Sporadic occurrence: Most cases of Goldenhar syndrome occur sporadically, meaning they arise due to random mutations during foetal development. The syndrome can affect individuals with no family history of the condition.

Goldenhar syndrome is a complex condition, and while the exact cause can be unclear in many cases, understanding the genetic and environmental influences may help guide treatment and management.

Testing & Diagnosis

The diagnosis of Goldenhar syndrome typically involves a combination of clinical evaluation, genetic testing, and imaging studies to confirm the presence of characteristic features and rule out other conditions with similar symptoms.

1. Clinical Evaluation

Physical examination: The initial diagnosis is often made through a detailed physical examination, during which a doctor will look for the typical characteristics of Goldenhar syndrome, such as:

- Facial asymmetry (hemifacial microsomia): One side of the face may be underdeveloped or smaller than the other, with differences in the development of the jaw, cheekbones, or eyelids.
- Ear anomalies: Underdeveloped or missing ears (microtia or anotia) and ear canal differences.
- Eye anomalies: Colobomas (a gap or a notch in one of the structures of the eye) or other eye differences, such as epibulbar dermoids (benign tumours on the eye surface).
- Vertebral anomalies: Some individuals may have irregularities of the spine, such as fusion of vertebrae (spinal fusion or scoliosis).
- Soft tissue anomalies: These may include cleft lip/palate, skin tags near the ears, or other skin-related features.

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- Family history: In some cases, the doctor may ask about any family history of similar features, although Goldenhar syndrome is more commonly sporadic (occurs randomly, without a family history).

2. Genetic Testing

Genetic testing is not always necessary for the diagnosis but may be used to confirm the condition. The genetic basis of Goldenhar syndrome is not fully understood, but it is thought to be associated with mutations in multiple genes, though no single gene has been definitively identified in all cases. Genetic testing may involve:

- Chromosomal analysis (karyotyping): This may be used to detect chromosomal differences, such as deletions, duplications, or other structural changes, although these differences are not present in all cases.
- Next-generation sequencing: This may help identify mutations in specific genes that are suspected to be involved in the syndrome, although the exact genetic causes are still being researched.

3. Imaging Studies

Imaging techniques help evaluate the extent of physical differences, especially in the skull, spine, and internal organs.

- X-rays: To assess vertebral anomalies (such as fused or misshapen vertebrae) and any skeletal irregularities.
- CT scan (Computed Tomography): A CT scan may be performed in order to get detailed images of any skull or craniofacial differences, especially if craniofacial differences like hemifacial microsomia or cleft palate are suspected.
- MRI (Magnetic Resonance Imaging): An MRI can be used to evaluate the brain, spinal cord, and soft tissue structures, as it provides a more detailed view than a CT scan.
- Ultrasound: In some cases, an ultrasound may be used to assess any internal organs, especially the kidneys and heart, as some individuals with Goldenhar syndrome may have related anomalies.

4. Hearing Tests

Since ear anomalies such as microtia (underdeveloped ear) and atresia (absence of ear canal) are common in Goldenhar syndrome, individuals may experience hearing loss. Therefore, an audiological evaluation is often recommended to assess the extent of hearing impairment. This could include:

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- Pure-tone audiometry: To assess hearing sensitivity at different frequencies.
- Speech audiometry: To determine the ability to hear and understand speech.
- Tympanometry: To evaluate the functioning of the middle ear.

5. Eye Examination

Because some individuals with Goldenhar syndrome may have colobomas or other eye anomalies, a comprehensive eye examination by an ophthalmologist is often recommended. The eye exam can help identify:

- Colobomas: Gaps or notches in the eye structures (such as the iris, retina, or optic disc).
- Epibulbar dermoids: Benign tumors that form on the surface of the eye.

6. Multidisciplinary Approach

Given the variety of potential complications in Goldenhar syndrome (including craniofacial, spinal, auditory, and visual difficulties), a multidisciplinary team may be involved in diagnosis and management of the condition. This may include:

- Paediatricians
- Genetic counsellors
- Otolaryngologists (ear, nose, and throat specialists)
- Ophthalmologists
- Orthopaedic surgeons
- Speech and developmental therapists

While the diagnosis of Goldenhar syndrome is often made based on an assessment by a doctor, genetic and imaging tests can help confirm the condition and assess its severity.

Treatment

The treatment of Goldenhar syndrome is largely symptomatic and supportive, as there is no cure for the condition. The treatment plan will depend on the severity of the symptoms and any associated medical issues. The goal of treatment is to manage and address differences, improve quality of life and address any complications that may arise.

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A multidisciplinary approach is typically required, as Goldenhar syndrome can affect multiple systems in the body, including the craniofacial structures, spine, ears, and eyes.

1. Craniofacial and Skull differences

Surgical intervention: Many children with Goldenhar syndrome have craniofacial problems such as hemifacial microsomia, craniofacial asymmetry, or cleft lip/palate. Surgical treatment can help address these issues and address appearance and improve function. Some potential procedures include:

- Craniofacial reconstruction: To address differences in the skull, jaw, and facial bones. Surgery may be done to reshape the facial bones and improve symmetry.
- Cleft lip/palate repair: If the individual has a cleft lip or palate, surgical repair is typically performed to improve speech and feeding ability.
- Ear reconstruction: Children with microtia (underdeveloped or absent ear) may require reconstructive surgery to form a more typical outer ear, which can also help with hearing function.

2. Hearing Loss

- Hearing aids: If the child has hearing loss due to anomalies in the ear or ear canal, hearing aids can help improve hearing.
- Surgical treatment: For those with atresia (absence of ear canal), surgery to create or reconstruct the ear canal and improve hearing may be considered. This may involve a procedure known as aural atresia repair.
- Speech therapy: If hearing loss affects speech development, speech therapy can be very beneficial in helping the child develop communication skills.

3. Spinal and Skeletal Anomalies

- Spinal surgery: Children with spinal irregularities, such as scoliosis or vertebral fusion, may require surgical intervention or a brace to address or stabilize the spine.
- Orthopaedic management: For those with limb anomalies (such as polydactyly or syndactyly), surgical treatment may be required to improve functionality and appearance. This could involve the removal of extra digits or separation of fused fingers.

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4. Eye Anomalies

- Surgical treatment: If the child has colobomas (a variation in eye formation where a gap or notch occurs in structures like the iris, retina, or optic nerve), surgical procedures may be needed in certain cases to address visual concerns or prevent complications.
- Vision therapy: Regular eye exams by an ophthalmologist are essential to monitor vision and detect any problems early. Corrective lenses may be prescribed if there are issues with refractive errors or other visual problems.

5. Developmental and Speech Therapy

- Speech therapy: Many children with Goldenhar syndrome may experience delays in speech development, especially if they have hearing loss, cleft palate, or other craniofacial issues. Speech therapy can help improve language skills.
- Developmental support: Children with developmental delays may benefit from early intervention programs that focus on motor skills, cognitive development, and social skills.

6. Psychosocial Support

- Psychological support: Given the physical and emotional challenges that may accompany Goldenhar syndrome, psychological counselling can be helpful for both the child and their family. This can assist with coping strategies, self-esteem, and adjustment to the condition.
- Social support: Support groups for families dealing with craniofacial conditions can offer valuable emotional support, resources, and information.

7. Monitoring and Follow-Up Care

Regular follow-up visits with specialists are essential to monitor for any complications or new issues that may arise. These specialists may include:

- Otolaryngologists (ENT specialists) for hearing and ear concerns.
- Ophthalmologists for eye monitoring and correction.
- Orthopaedic surgeons for skeletal and spinal issues.
- Speech therapists and developmental specialists for growth and speech issues.

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Did you know?

In Australia, approximately 1 baby in 10,000 is diagnosed with Goldenhar syndrome.*

Goldenhar syndrome is also known as "oculoauriculovertebral spectrum" (OAVS) because it affects the development of the eyes (oculo-), ears (auriculo-), and spine (vertebral).

References:

*Junaid, M., Slack-Smith, L., Wong, K., Bourke, J., Baynam, G., Calache, H., & Leonard, H. (2022). Epidemiology of Rare Craniofacial Anomalies: Retrospective Western Australian Population Data Linkage Study. The Journal of Pediatrics, 241, 162-172.e169 https://doi.org/10.1016/j.jpeds.2021.09.060

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How we can support you:

- Care packs
- Financial assistance
- Family support coordinator
- Connection to other families