

Goldenhar Syndrome

Also known as: facioauriculovertebral (FAV) syndrome, oculoauriculovertebral dysplasia, craniofacial microsomia, lateral facial dysplasia

Clinical Characteristics

Goldenhar syndrome primarily involves malformations of the cheekbones, jaw, mouth, ears, eyes, and vertebrae (in other words, first and second branchial arch derivatives). Craniofacial abnormalities include malar, maxillary, mandibular and temporal hypoplasia, macrostomia, underdeveloped facial muscles, cleft lip and/or cleft palate, abnormalities of the teeth, epibulbar dermoids and lipodermoids, coloboma, microphthalmia, strabismus, and ear abnormalities with or without hearing loss. The vertebrae may be absent, incompletely developed, or fused. Occasionally, there may be skeletal, neurological, cardiac, pulmonary, renal, and/or gastrointestinal issues.

Goldenhar Syndrome and Hearing Loss

Ear abnormalities include microtia or anotia, atresia of the ear canals, preauricular tags, and middle/inner ear abnormalities. Hearing loss may be conductive or sensorineural, conductive loss being more common due to malformations of the middle or outer ear structures. The loss may be unilateral or bilateral. Amplification is often an issue because of ear malformations. It appears that most patients have a non-progressive form of hearing loss.

Natural History

Goldenhar syndrome is diagnosed in both males and females, though is slightly more common in males (3:2 ratio). It can also be found in all ethnicities. The estimated frequency of the syndrome is about 1 in 5600. Goldenhar syndrome can be considered part of the oculoauriculovertebral spectrum (OAVS), or the facio-auriculo-vertebral spectrum. It is the "more severe" of two manifestations of OAVS, the other being Hemifacial microsomia. The major difference between the two is that Hemifacial microsomia is mostly characterized by abnormalities of the jaw, while Goldenhar syndrome affects distal structures like the eyes and spine in addition to the mandible and maxilla. Both Hemifacial microsomia and Goldenhar syndrome are more likely to occur unilaterally (the right side being the more commonly affected side), but of the two, Goldenhar syndrome is slightly more likely to show bilateral manifestations.

The characteristics of Goldenhar syndrome are present at birth and are generally non-progressive. The greatest issues facing patients at diagnosis involve breathing and feeding, and these must be addressed in a timely fashion. Mild mental retardation is present in about 5-15% of patients, but most have normal intelligence. A normal life span is expected.

Genetics

Although rare cases of autosomal dominant, autosomal recessive, and X-linked inheritance patterns have been reported in the literature, Goldenhar syndrome is considered a sporadic disorder. Possible “causes” of the condition include chromosomal abnormalities, teratogenic drugs, hyperglycemia, genomic imprinting or mosaicism, sporadic mutations, and vascular disruptions during pregnancy. Recurrence risks of 1% and 3% are estimated for siblings and offspring of affected individuals, respectively.

Management

The diagnosis of Goldenhar syndrome is made clinically. Currently there is no genetic testing available on a clinical basis, though research testing is being performed.

Initial evaluations should include those for adequacy of breathing, swallowing and feeding, hearing, and vision. Further evaluations for gastroenterology and cardiology should be performed at some point. A spinal assessment, including appropriate imaging, is indicated. Management should be handled by a multidisciplinary craniofacial team, including a medical geneticist, surgeon, audiologist, and psychologist. Speech and occupational therapy may be needed. In the early years, proper positioning of the airway and a gastrostomy tube could be needed. Reconstructive surgery is standard procedure and should be scheduled as needed.

Many individuals with Goldenhar syndrome face social challenges as they grow older. Children and adolescents may face teasing or rejection due to their appearance. These issues must be handled sensitively and openly. Many people find that talking to a counselor or psychologist helpful, while others find their own coping strategies. This should be discussed with parents at length while their child is still young so they may be prepared for such issues in the future.

Resources for Families

Statewide Genetics Program

Phone: 608-267-7148

Fax: 608-267-3824

Email: meyeram@dhfs.state.wi.us

Wisconsin First Step Hotline

Phone: 1-800-642-7837 voice/TTY

Website: www.mch-hotlines.org

Wisconsin Office for Deaf and Hard of Hearing

Phone: 1-608-266-3118 voice/TTY

Website: www.dhfs.state.wi.us/sensory

Regional Children and Youth with Special Health Care Needs Centers

Centers in Green Bay, Wausau, Milwaukee, Madison, and Chippewa Falls

Website: http://dfhs.wisconsin.gov/DPH_BFCH/cshcn/index.HTM

WI Chapter of Families for Hands & Voices

Phone: (920) 437-7370

Website: www.handsandvoices.org

Parent-to-Parent of Wisconsin

Phone: 1-888-266-0028

Email: rmathea@shsmh.org

Family Village online resource

Library Card Catalog of Disorders

www.familyvillage.wisc.edu

Goldenhar Syndrome Support Network Society

9325 163rd St.

Edmonton, Alberta

T5R 2P4 Canada

Email: support@goldenharsyndrome.org

Website: www.goldenharsyndrome.org

National Organization for Rare Disorders (NORD)

www.rarediseases.org