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**Goldenhar's Syndrome: Characteristics,
Audiological Management, and
Developmental Outcomes**

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Physical and Medical Characteristics

Definition

- The first report of a child bearing the characteristics of Goldenhar's syndrome may have been made by von Arlt in 1845.
- However, Goldenhar is credited with reviewing the literature and reporting three new cases of patients exhibiting this particular collection of congenital abnormalities in 1952 (Gorlin, Jue, Jacobsen, & Goldschmidt, 1963).
- Goldenhar described epibulbar dermoid cysts and preauricular skin accessories as the hallmarks of the syndrome that now carries his name (Bayraktar, Bayraktar, Ataoglu, Ayaz, & Elevli, 2005; Strömmland et al., 2007).
- In 1963, Gorlin and colleagues coined the term oculoauriculovertebral dysplasia after adding vertebral anomalies to Goldenhar's list of symptoms (Gorlin et al., 1963).
- Several expressions are used today to describe Goldenhar's syndrome, a disorder which remains nebulous and somewhat poorly defined. Other terms that are used to describe the syndrome are **oculoauriculovertebral spectrum (OAVS)** and **hemifacial microsomia (HFM)** (Araneta et al., 1997; Cousley & Calvert, 1997).
- Some researchers argue that Goldenhar's syndrome is a widely recognized label, although OAVS is more precise because it describes the typical anomalies associated with the disorder. Goldenhar's syndrome may be considered a specific clinical profile that is included under the broader category of OAVS, and HFM

has been described as a spectrum that includes both OAVS and Goldenhar's syndrome (Araneta et al., 1997; Cousley & Calvert, 1997; Strömmland et al., 2007).

- In the literature, these terms, particularly Goldenhar's syndrome and OAVS, appear to be interchangeable (Araneta et al., 1997). The expression Goldenhar's syndrome will be used in this paper to refer to all manifestations of the disorder, although several works cited may use other terms in their descriptions. When working with families of children who have Goldenhar's syndrome, professionals may choose to inform the parents that these other labels exist. This information may be important to promote parent understanding, research, and self-advocacy.

Physical Anomalies

- Four major characteristics are included in Goldenhar's syndrome:
 - eye anomalies,
 - ear anomalies,
 - vertebral anomalies, and
 - facial asymmetry (Bayraktar et al., 2005; Cousley & Calvert, 1997; Strömmland et al., 2007; Tasse et al., 2005).
 - The syndrome is highly variable and may display many other complications (Bisdas, Lenarz, Lenarz, & Becker, 2005; Cousley & Calvert, 1997; Strömmland et al., 2007; Tasse et al., 2005).
 - Abnormalities tend to be unilateral, although they may sometimes appear bilaterally (Strömmland et al., 2007).
 - Specific manifestations of Goldenhar's syndrome may include the following, although this list is far from complete:

- Eye anomalies: epibulbar dermoids, colobomas of the upper lids (Araneta et al., 1997; R. Wang, Martínez-Frías, & Graham, 2002).
- Ear anomalies: preauricular tags and pits, microtia, atresia, ossicular chain dysplasia,
- enlarged bony portion of the eustachian tube, inner ear malformations (common cavity,
- enlarged vestibule,
- enlarged internal auditory canal,
- absent or dysplastic semicircular canals, absence of cochlea) (Bisdas et al., 2005; Haginomori et al., 2003; Lemmerling, Vanzielegem, Mortier, Dhooge, & Kunnen, 2000; Scholtz et al., 2001; Strömmland et al., 2007; R. Wang et al., 2002).
- Vertebral anomalies: thoracic hemivertebrae anomalies (R. Wang et al., 2002).
- Facial anomalies: Malar and/or mandibular hypoplasia, cleft lip or palate, tracheoesophageal fistula (Araneta et al., 1997; R. Wang et al., 2002).
- Central nervous system anomalies: encephalocele, holoprosencephaly, Arnold-Chiari malformation, lymphomas in the corpus callosum, hydrocephalus, lissencephaly (Araneta et al., 1997; Bayraktar et al., 2005; Johansson et al., 2007; Strömmland et al., 2007).

- Other: renal anomalies, heart anomalies (ventricular septal defect, tetralogy of Fallot) (Araneta et al., 1997; Bayraktar et al., 2005).

Diagnosis

- Professionals in pediatric medicine, radiology, ophthalmology, otolaryngology, odontology, and neurology contribute to the diagnosis of Goldenhar's syndrome through identification of physical anomalies (Araneta et al., 1997; Strömmland et al., 2007; Tasse et al., 2005).
- Due to the congenital nature of Goldenhar's syndrome, the most apparent physical anomalies are obvious at birth.
- Certain techniques, such as fetal sonographic biometry and 3D sonography, may allow prenatal diagnosis to be possible in some cases (Martinelli et al., 2004; Volpe & Gentile, 2004).
- The diagnosis of Goldenhar's syndrome may be complicated by the lack of any established diagnostic criteria, although ear anomalies are generally an essential component of the diagnosis.
- In fact, some clinicians consider microtia to be a minimal manifestation of Goldenhar's syndrome, and some diagnoses may be made based on isolated microtia alone (Araneta et al., 1997; Llano-Rivas, González-del Angel, del Castillo, Reyes, & Carnevale, 1999).
- Goldenhar's syndrome is classified in the International Statistical Classification of Diseases and Related Health Problems, 10th Revision, under the category of "congenital malformation syndromes predominantly affecting facial appearance" (ICD-10 code: Q87.0) (WHO, 2006).

- Although this classification exists, Goldenhar's syndrome has no generally accepted system of scoring for diagnosis or severity. Tasse et al. (2005) proposed a classification system based on the presence of the main clinical characteristics and additional clinical findings as well as the appearance of these anomalies as unilateral or bilateral.
- If used more widely, such a system could help provide further definition to the understanding of Goldenhar's syndrome for families and clinicians.

Hearing Loss and Sensory Characteristics

- Children with Goldenhar's syndrome usually exhibit conductive hearing loss due to malformations of the conductive mechanism of the ear (Cousley & Calvert, 1997; Scholtz et al., 2001; R. Wang et al., 2002).
- External ear and middle ear anomalies are commonly present and well documented in children with Goldenhar's syndrome (Bisdas et al., 2005; Cousley & Calvert, 1997).
- In one study, 12 out of 14 children exhibited atresia of the external auditory canal, and the ossicular chain was dysplastic in 14 and absent in one of 28 ears (Bisdas et al., 2005).
- Haginomori et al. (2003) reviewed a case study of a child who exhibited an enlarged bony portion of the eustachian tube as well as an ossicular anomaly.
- In another study, 17 out of 18 patients displayed external ear anomalies, and 12 out of 18 patients showed anomalies of the middle ear (Strömmland et al., 2007).
- These physical abnormalities of the ear generally result in conductive hearing loss.

- Reports of children with Goldenhar's syndrome show that a conductive component to hearing loss is present in about two-thirds of patients (D'Antonio, Rice, & Fink, 1998; Strömmland et al., 2007).
- Congenital conductive hearing loss from conditions like Goldenhar's syndrome usually falls in the mild to moderate range, with patients showing about 40-60 dB of hearing loss (Stewart & Downs, 1993; Van Meter & Weaver, 1996).
- Due to the heterogeneity of Goldenhar's syndrome, not all children exhibit conductive hearing loss.
- Some patients will display sensorineural hearing loss or mixed hearing loss resulting from various abnormalities.
- Anomalies of the inner ear occur in some children with Goldenhar's syndrome, although they tend to be rare (Lemmerling et al., 2000; Scholtz et al., 2001).
- Many of these abnormalities occur in the vestibular system, including enlarged or underdeveloped vestibule and absent or dysplastic semicircular canals (Bisdas et al., 2005; Haginomori et al., 2003; Strömmland et al., 2007).
- Other inner ear anomalies that have been reported are common cavity and absence of the cochlea (Bisdas et al., 2005; Haginomori et al., 2003).
- Inner ear anomalies may result in sensorineural hearing loss in some cases. Cousley and Calvert (1997) report that 6-16 % of patients with Goldenhar's syndrome display sensorineural hearing loss.
- Other reports range from 22% of patients with either sensorineural or mixed hearing loss (D'Antonio et al., 1998) to one-third of patients having a sensorineural component (Strömmland et al., 2007).

- Interestingly, ear anomalies and hearing loss tend to present bilaterally in Goldenhar's syndrome, whereas facial effects are usually unilateral (Bayraktar et al., 2005; Strömmland et al., 2007).

Visual Impairment:

- While the focus of this paper is on hearing loss and its effects, it is important to note that children with Goldenhar's syndrome may experience visual impairment in addition to or in isolation from hearing loss.
- In one study, 5 out of 16 patients exhibited visual impairment, with two of them being blind and one being deaf-blind (Strömmland et al., 2007).
- Additional sensory impairments may complicate the management of children with hearing loss and should be considered in the assessment, treatment, and counseling of these patients.

Developmental Characteristics

- Many patients with Goldenhar's syndrome will have normal intelligence and development unless other disorders are present in combination (R. Wang et al., 2002).
- However, mental retardation and developmental delays may be associated in an estimated 5-15% of cases (Bayraktar et al., 2005; Scholtz et al., 2001; Strömmland et al., 2007; Tasse et al., 2005).
- Johansson et al. (2007) report some estimates of the occurrence of learning disabilities in Goldenhar's syndrome of up to 45%, and they report learning

disabilities in 82% of patients with central nervous system (CNS) abnormalities and Goldenhar's syndrome.

- Recently, investigators have discovered a greater prevalence of autism and autistic-like features in children with Goldenhar's syndrome than in the general population (Strömmland et al., 2007).
- One study of 20 subjects with Goldenhar's syndrome revealed autism spectrum disorders (ASD) in 42% of patients and full autism in 11% of patients;
- in contrast, the prevalence of ASD in the general population is .5%-1% and the prevalence of autism is .1-.2% (Johansson et al., 2007).
- As with sensory impairments, any developmental delays or psychosocial disorders must be considered in the management of children with Goldenhar's syndrome.

Incidence

- Estimates of the incidence of Goldenhar's syndrome are inconsistent and vary widely among reports.
- This may be due in part to the heterogeneity of the disorder and its lack of definition in the field (Strömmland et al., 2007).
- Cases of Goldenhar's syndrome are usually sporadic (Bayraktar et al., 2005; Cousley & Calvert, 1997; Lemmerling et al., 2000; R. Wang et al., 2002).
- Familial transmission is rare, but may be seen in 1-2% of cases (Cousley & Calvert, 1997; Tasse et al., 2005; R. Wang et al., 2002).
- Some estimates of the incidence of Goldenhar's syndrome are as follows: 1 in 3000 to 1 in 5000 (Martinelli et al., 2004), 1 in 5000 (Johansson et al., 2007), 1 in 5600 (Bayraktar et al., 2005; Strömmland et al., 2007), 1 in 5000 to 1 in 25,000 (R.

Wang et al., 2002), 1 in 5000 to 1 in 45,000 (Tasse et al., 2005), and 1 in 45,000 in Northern Ireland (Scholtz et al., 2001).

- At the University of Colorado Hospital in Denver, Colorado, one infant out of 565 infants with hearing loss was diagnosed with Goldenhar's syndrome from the years 1964-1991 (Stewart & Downs, 1993).
- Goldenhar's syndrome is seen more often in males than in females (about 3:2 male to female), and the recurrence risk is about 2-6% (Bayraktar et al., 2005; Johansson et al., 2007; Martinelli et al., 2004).
- In autosomal dominant Goldenhar's syndrome, patients more often show bilateral effects and less often show hearing loss when compared with sporadic cases (Tasse et al., 2007).

Causes and Risk Factors

- Goldenhar's syndrome arises from a developmental irregularity of the first and second branchial arches (Araneta et al., 1997; Bisdas et al., 2005; Cousley & Calvert, 1997; R. Wang et al., 2002).
- This disruption in development appears to occur at either the fourth week of gestation or from about 30-45 days of gestation (Bisdas et al., 2005; Tasse et al., 2005).
- The exact reason for the developmental disturbance varies among cases. Teratogens such as retinoic acid, primidone, and thalidomide may interfere with

embryonic development (R.R. Wang & Andres, 1999), or there may be a chromosomal component to the disruption.

- Several risk factors exist for Goldenhar's syndrome.
- Infants of diabetic mothers show an increased risk of developing the disorder, as diabetes is thought to interfere with neural crest cell migration (Bisdas et al., 2005; R. Wang et al., 2002).
- In a study performed by Araneta and colleagues (1997), infants of Gulf War veterans displayed an incidence of 14.7 per 100,000 for Goldenhar's syndrome, whereas infants of veterans who were not deployed to the Gulf War showed an incidence of 4.8 per 100,000; however, this difference was not statistically significant.
- Several researchers have reported that there may be some connection between Goldenhar's syndrome and the use of assisted reproductive technology, although more research is needed in this area (Johansson et al., 2007; Strömmland et al., 2007).
- Additionally, mothers of children with Goldenhar's syndrome have reported other factors, such as early bleedings during pregnancy and smoking more than 10 cigarettes per day during pregnancy (Strömmland et al., 2007).
- Although research is still being performed regarding genetics and Goldenhar's syndrome, family history may be a possible risk factor for developing the disorder (Cousley & Calvert, 1997).

- Some relationships seem to exist between Goldenhar's syndrome and other congenital conditions, particularly between Goldenhar's syndrome and the CHARGE association.
- Anomalies often seen as features of the CHARGE association are choanal atresia, colobomata, ear anomalies, hearing loss, heart defects, genital hypoplasia, growth retardation, developmental delays, CNS anomalies, tracheoesophageal fistula, facial palsy, renal anomalies, and cleft lip and palate (Van Meter & Weaver, 1996).
- The overlap of characteristics of Goldenhar's syndrome and the CHARGE association shows a statistical relationship and may indicate a common pathogenic mechanism between the two disorders (Källén, Robert, Castilla, Mastroiacovo, & Källén, 2004; Van Meter & Weaver, 1996).
- Patients who show features of both Goldenhar's syndrome and the CHARGE association tend to be more severely affected than those who are characterized by one or the other of the disorders (Johansson et al., 2007).
- Clinicians should be aware of this possible association and the effects it may have.

Management and Interventions

Early Intervention and Treatment Planning

- Many bodily systems are implicated in Goldenhar's syndrome.

- Therefore, many professionals are needed to manage a child with the disorder. Treating a child with Goldenhar's syndrome requires long-term planning by a cooperative multi-disciplinary team.
- The goals of treatment are to attain the best possible aesthetics and functionality, including improved appearance, proper use of affected systems, appropriate speech and language, proper physical growth, and optimal cognitive, social, and emotional development (Belenchia & McCardle, 1985; Cousley & Calvert, 1997).
- Some disciplines that must be integrated into the management of a child with Goldenhar's syndrome are audiology, genetics, neurology, orthodontics, otolaryngology, psychology, reconstructive surgery, and speech-language pathology (Cousley & Calvert, 1997).
- As with other disorders that may affect development, early intervention is key in the treatment of children with Goldenhar's syndrome (Bayraktar et al., 2005; Belenchia & McCardle, 1985; Stewart & Downs, 1993).
- The development of age-appropriate language depends heavily on early and effective intervention. A family-centered approach to intervention can provide essential support to the parents of a child with Goldenhar's syndrome and encourage learning within the home (Belenchia & McCardle, 1985).
- Greatest efforts must be taken to properly identify children with Goldenhar's syndrome and enroll them immediately in early intervention programs.

Medical Intervention

- Depending on the severity of the abnormalities, children with Goldenhar's syndrome may need to go through extensive medical interventions.

- Some steps that may need to be taken include correction of abnormalities in soft tissue and craniofacial skeletal structures, repair of nerves, middle ear reconstruction, and the use of orthodontic appliances (Cousley & Calvert, 1997).
- Advances in tissue engineering and computer design have provided options for treatment such as improved ear prosthetics and reconstructive ear surgery.
- Medical interventions must follow a planned sequence depending on the patient's needs. In some cases, surgeons begin with the facial skeleton and then move to soft tissue reconstructions, and in other cases, the soft tissue abnormalities are addressed first (R.R. Wang & Andres, 1999).
- The timing of corrections involving skeletal structures is controversial among medical interventionists.
- Some practitioners believe that early treatment is better and may improve a child's growth and development.
- However, the temporal bones of children are thin and soft while in development, and performing skeletal reconstruction on a child may pose many risks.
- These risks and benefits to the timing of intervention must be considered when planning medical treatment for a child with Goldenhar's syndrome (Cousley & Calvert, 1997; R.R. Wang & Andres, 1999).
- In addition to the management of anomalous craniofacial structures, children with Goldenhar's syndrome may need intervention for heart abnormalities, vertebral anomalies, hydrocephalus, and many other conditions that could co-exist.
- The multidisciplinary management team must be aware of the treatments that are necessary for the overall health of the child and must make referrals accordingly.

Audiological Intervention

- Because most children with Goldenhar's syndrome have normal intelligence, audiological assessment should usually entail the same procedures as with a typically developing child.
- When secondary disabilities are present, the audiologist will need to adjust procedures accordingly.
- Children with Goldenhar's syndrome and other complications, such as autism or a cognitive delay, should be assessed according to their developmental age and abilities.
- In such cases, an audiologist may need to rely heavily on physiological assessments like auditory brainstem audiometry.
- However, some modifications may be made to obtain reliable behavioral results. For example, the audiologist should strive to make the task meaningful for the child, accept unconventional responses in visual reinforcement or play audiometry, use strong social reinforcement or tangible reinforcement when necessary, and attempt to make the child feel comfortable in the testing environment.
- For children with Goldenhar's syndrome and hearing loss, early amplification is of utmost importance in order to give the children access to sound at a young age (Bayraktar et al., 2005; Cousley & Calvert, 1997; Stewart & Downs, 1993; Strömmland et al., 2007).

- The type of technology that is appropriate for a particular child may be a hearing aid, bone-anchored hearing aid (BAHA), or cochlear implant, depending on that child's type and degree of hearing loss (Strömmland et al., 2007).
- Many patients with Goldenhar's syndrome have a congenital conductive hearing loss, and a BAHA is the treatment of choice for such patients (Stewart & Downs, 1993).
- Compared with traditional hearing aids, a BAHA can be good for retention, minimized skin irritation, and improved cosmetics (R.R. Wang & Andres, 1999). One study showed that out of 40 patients with conductive hearing loss and bone-conduction pure tone average levels of 60 dB or less, each patient showed an improvement of 32 +/- 19 dB with the use of a BAHA (Lustig et al., 2001).
- Additionally, children with Goldenhar's syndrome, conductive hearing loss, and developmental delays may be considered good candidates for BAHA's. Although patients with mental retardation had previously been excluded from candidacy for BAHA's due to concerns regarding their ability to care for the devices, current research shows that patients with mild or moderate mental retardation may be good candidates for BAHA's in the case of conductive hearing loss (Kunst et al., 2006).
- Children with Goldenhar's syndrome who have sensorineural or mixed hearing loss need to be managed differently from children with purely conductive hearing loss.

- Research investigating the treatment of children with craniofacial abnormalities and sensorineural hearing loss is lacking, yet clinicians may use related ideas and professional experience to develop proper management procedures.
- While conductive hearing loss results in diminished audibility of sounds, sensorineural hearing loss causes diminished audibility as well as compromised resolution of sounds. Even with proper amplification, children with sensorineural hearing loss may not hear with the same clarity as a person with normal hearing.
- Therefore, these children will likely need different intervention strategies from their peers with conductive hearing loss.
- A child with sensorineural hearing loss may need a greater extent of auditory training and speech and language therapy than a child with conductive hearing loss.
- Audiologists and speech-language pathologists should collaborate and share information with one another to provide the best possible management for children with Goldenhar's syndrome.
- Professionals and caregivers must be aware of the differences that may exist among children with this heterogeneous disorder and plan a child's intervention based on his or her individual needs.

Speech and Language Intervention

- An area in which early intervention is absolutely crucial for optimal outcomes is speech and language development. As with other disciplines, the research specifically relating to speech and language intervention for children with Goldenhar's syndrome is minimal.

- However, a small amount of research has been performed in this area. According to a case study by Belenchia & McCardle (1985), children with normal intelligence and Goldenhar's syndrome may be expected to show substantial progress if speech and language intervention is provided early.
- A broader study investigating conductive hearing loss resulting from many etiologies indicated that children with conductive hearing loss and normal cognitive development can be expected to have normal speech and language development if they are provided with early amplification and appropriate therapy (Stewart & Downs, 1993).
- Interventionists may enter treatment with high expectations, but it should be remembered that in Goldenhar's syndrome, abnormal craniofacial structures may interfere with typical speech production, and articulation therapy may be necessary (Belenchia & McCardle, 1985).
- When secondary disabilities are present, clinicians must adapt their assessment and intervention strategies to accommodate for such issues. Children who have Goldenhar's syndrome and disorders such as autism may need alternative communication systems, educational approaches, and behavioral interventions (Strömmland et al., 2007). Interventionists must use their expertise to treat the effects of hearing loss, oro-facial abnormalities, and any developmental or cognitive differences and promote the development of speech and language in children with Goldenhar's syndrome.

Parent Factors and Counseling

- A significant component of intervention when managing a child with Goldenhar's syndrome relates to counseling of the child and family.
- Parents of children with Goldenhar's syndrome may experience feelings of shock, grief, or disbelief upon discovering their child's condition. They may feel worried or distressed about many factors relating to the syndrome. Parents may be shocked and disturbed by the physical appearance of their child, they may be concerned about the child's mental abilities, they may worry for their child's emotional well being, or they may fear physical harm or death for their child (Endriga & Kapp-Simon, 1999). All of these feelings may need to be addressed at some point by the multidisciplinary management team.
- As children with Goldenhar's syndrome grow older, parents may feel greater concern over their children's social behaviors and adjustment. In a collection of interviews conducted to understand mothers' perceptions in raising children with craniofacial anomalies, mothers reported protectiveness toward their children. They were concerned for their children's physical, social, and emotional well-being. Mothers reported that their children's strengths included determination, perseverance, and sociability. They described proactive parenting as encouraging independence and peer interactions in their children (Klein, Pope, Getahun, & Thompson, 2006).
 - Some examples of these responses are included below:
 - When she has her . . . soccer practice . . . I'm the only parent that stays the whole time . . . Just in case something happens, I'm there. I'm a little more overprotective with her . . . Oh, if she ever

gets hit in the face with something, I would die, I think . . . To have to go through mouth surgery again (p.592).

- A trip to the ball game. Usually it's really fun, and he has a great time . . . But . . . you could get someone really mean two rows away from you and some people are really . . . cruel. I'm not talking little kids who don't know what they're saying . . . I mean adults (p.592).
- When she wanted to learn how to do the monkey bars. Her physical therapist called us and told us [the child] really wants to do this. So every day, for maybe 6 weeks to 2 months . . . she and my husband went to the park and did the hand-over-hand thing (p. 593).
- All of these concerns, perceptions, and behaviors must be addressed by the management team. While a professional in psychology should be involved to help support the family, many of these concerns may also be addressed by other members of the team, such as the pediatrician, audiologist, and speech-language pathologist. Members of the multidisciplinary intervention team should discuss the overall well-being of the family and share information and strategies to help support the parents and child.

Behavioral Characteristics and Outcomes

Auditory Perception and Skills

- Clinicians may expect many children with Goldenhar's syndrome to have abnormal auditory perception and auditory skills due to the likely presence of conductive, sensorineural, or mixed hearing loss.
- Although research in Goldenhar's syndrome has focused primarily on medical aspects of the disorder, practitioners may make assumptions and form decisions based on the evidence that is available in related areas and the use of professional judgment.
- In one case study reported in 1985, a one-year-old boy with Goldenhar's syndrome responded to his name at 15 dB HL, a 1500-Hz warble tone at 20 dB HL, and a bell at 30 dB HL (Belenchia & McCardle, 1985).
 - These responses from sound field stimuli are thought to represent the boy's left ear due to atresia of his right ear canal.
 - From these results, a reader would expect the boy's sensorineural mechanism to be intact, an expectation that is consistent with what has been shown in the medical literature.
 - However, a good clinician will recognize that this is one example of one child among many children with heterogeneous symptoms, and the information in this report is not complete.
 - Additionally, the case study is over 20 years old, and many advances in auditory assessment have been made during that time.
- Other evidence must be assessed in order to form expectations of auditory behaviors in children with Goldenhar's syndrome.

- Stewart and Downs (1993) explained that low frequencies are often more affected than high frequencies in a conductive hearing loss.
- Conversely, clinical experience will show that high-frequency loss is quite common in sensorineural hearing loss, with better hearing appearing in the low frequencies.
- These differences may produce distinct outcomes for speech understanding based on a child's specific array of anomalies.
- For instance, a child with conductive hearing loss may have difficulty distinguishing some vowels, which carry their greatest energy in the low frequencies;
- however, a child with high-frequency sensorineural hearing loss may show more difficulty distinguishing consonants and certain vowels with similar high-frequency formants.
- Both of these issues may be alleviated by the use of proper amplification. Each child will have different strengths and needs in their ability to perceive and understand sounds.
- These abilities must be assessed through various instruments, such as the Functional Auditory Performance Indicators (FAPI) by Stredler-Brown and Johnson (2004), and development of these skills must be addressed in therapy. Auditory skills should not only be developed when the child is working with a clinician, but strategies should be given to parents to encourage auditory skill development in the home.

Speech

- Several factors may affect outcomes in the speech of children with Goldenhar's syndrome, including hearing loss, oro-facial abnormalities, and cognitive or neuropsychiatric disorders (Strömmland et al., 2007).
- For children with conductive hearing loss only, the expectation for speech is quite good (Stewart & Downs, 1993).
- However, the presence of craniofacial anomalies may complicate the ability of a child to develop perfectly normal speech.
- These structural abnormalities may create difficulties in articulation, and laryngeal anomalies may impact a child's airway function, voice quality, and speech intelligibility (Belenchia & McCardle, 1985; D'Antonio et al., 1998).
- In one case study, a young boy with Goldenhar's syndrome exhibited hypernasality with audible nasal emission (Belenchia & McCardle, 1985).
- In another study, eight out of 18 patients showed speech problems (Strömmland et al., 2007).
- A study by D'Antonio, Rice, and Fink (1998) yielded the following results: out of 41 patients, 39% showed hypernasality and 42% showed hoarseness; out of 18-20 patients, 74% showed impaired articulation, 32% showed asymmetric facial motion and nasal grimacing, 56% showed impaired intelligibility, 16% displayed hyponasality, 22% showed borderline or inadequate velopharyngeal function, 35% showed harsh voice quality, 10% showed high-pitched voice, 5% showed breathy voice quality, and 15% showed moderate or severe laryngeal voice symptoms.

- The above symptoms are examples of abnormalities that some children with Goldenhar's syndrome may exhibit in their speech. Parents should be prepared that their child with Goldenhar's syndrome may struggle with speech due to craniofacial anomalies, and appropriate therapy must be provided. Speech-language pathologists should work with the otolaryngologist and reconstructive surgeon on the management team to monitor the child's progress as well as any medical interventions that may affect speech.

Language

- Language development in children with Goldenhar's syndrome depends on several factors.
 - Hearing loss, whether conductive or sensorineural in nature, may affect language development.
 - For example, Belenchia & McCardle (1985) reported significant delays in overall language development in their case study of a one-year-old boy with Goldenhar's syndrome.
 - However, if hearing loss is identified and treated early, outcomes should be quite positive for the development of appropriate language (Yoshinaga-Itano, Sedey, Coulter, & Mehl, 1998).
 - After four months of language therapy, the young boy in the Belenchia & McCardle (1985) study performed above age level for receptive and expressive language, auditory comprehension, and verbal ability.
- As with other issues discussed in this paper, the research investigating language development in children with Goldenhar's syndrome is sparse.

- Clinicians may expect that other factors aside from hearing loss, such as cognitive abilities or the presence of neuropsychiatric disorders, may affect language development in children with Goldenhar's syndrome.
- In the best case scenario, clinicians may expect that children with conductive hearing loss and normal intelligence should have normal language development with the proper amplification and intervention.
- The boy in the case study discussed above had conductive hearing loss and normal intelligence, and he was able to excel in language after only four months of intervention (Belenchia & McCardle, 1985).
- However, the presence of sensorineural hearing loss or any complicating disorders may negatively impact language development.
- A child with Goldenhar's syndrome, middle and inner ear anomalies, and a cognitive delay may need extensive intervention to develop age-appropriate language.

Developmental Outcomes

- Developmental outcomes for children with Goldenhar's syndrome may vary greatly depending on their specific presentation of physical anomalies and the presence of secondary disabilities.
- Young infants with Goldenhar's syndrome may experience difficulties in feeding due to problems in their general health (heart defects, breathing or gastrointestinal problems) or due to their craniofacial anomalies (Endriga & Kapp-Simon, 1999; Strömland et al., 2007).

- Infants may also display late walking if vestibular anomalies are present (Strömmland et al., 2007).
- In case studies conducted by Van Meter and Weaver (1996), reporting on two children with Goldenhar's syndrome, both children exhibited delays in gross motor skills (one child at 17 months with skills at 13 months, and one child at 9 months with skills at 5 months), and one child showed delays in adaptive/fine motor skills (at 17 months with skills at 13 months and 15 months).
- These are isolated examples of possible developmental outcomes in children with Goldenhar's syndrome.
- Developmental assessments must be included in the evaluation of children with Goldenhar's syndrome. The results of these assessments may assist in establishing expectations and treatment protocols for the child.
- Depending on the child's abilities, modifications may need to be made to the assessment procedure.
- Clinicians should be careful to select instruments that emphasize the child's strengths while recognizing areas of need.
- The results of developmental assessments may be helpful to professionals in audiology, speech-language pathology, neurology, psychology, and several medical realms, and they should be shared among these members of the management team to promote optimal understanding and treatment for the child.

Social-Emotional Outcomes

- Several factors may contribute to the social-emotional characteristics of children with Goldenhar's syndrome.
- The craniofacial anomalies associated with Goldenhar's syndrome may psychologically impact the child (R.R. Wang & Andres, 1999).
- Reports on the social and emotional functioning of children with craniofacial syndromes are varied.
- Some behaviors that may be exhibited by these children are social withdrawal, isolation, and internalizing or externalizing behaviors.
- Some studies show that children with craniofacial anomalies have self-concepts in the normal range, and some report that these children show lower self-concepts than the general population.
- Although most children with craniofacial anomalies function normally in the social-emotional realm, there are more children in this population who show behavioral and emotional difficulties than in the population of their typically developing peers (Endriga & Kapp-Simon, 1999).
- The social-emotional outcomes of children with Goldenhar's syndrome may be closely related to the adjustment and functioning of their parents.
- Research shows that parent factors can predict the adjustment of children with craniofacial anomalies to a great extent. For instance, parental stress may lead to poorer emotional health of children with craniofacial anomalies as well as less confident parenting and even reduced marital satisfaction. Parental stress may also affect parent-child interactions,

although these results are mixed. Higher maternal stress during preschool years predicts the social-emotional outcomes of the child in elementary school, including poorer social competence and increased behavior problems (Endriga & Kapp-Simon, 1999).

- A child's social-emotional functioning may be expected to be related to the medical severity of the craniofacial anomalies that are present; however, maternal adjustment and maternal perceptions of the mother-child relationship are more closely linked to a child's emotional adjustment than medical severity (Campis, Demaso, & Twente, 1995).
- Certain measures may be taken to reduce parental stress and improve social-emotional outcomes in children with Goldenhar's syndrome.
- Higher levels of social support are related to better adjustment for both the parent and child (Campis et al., 1995).
- Interventionists may encourage social support by introducing families of children with craniofacial anomalies to one another.
- Additionally, positive role models could be provided who have lived with Goldenhar's syndrome or other craniofacial anomalies.
- Another possible strategy to reduce parental stress is the provision of early intervention (Pipp-Siegel, Sedey, & Yoshinaga-Itano, 2002). Parents may find the extra support of early intervention to alleviate some of the pressures and questions that accompany Goldenhar's syndrome and similar congenital conditions. With quality early intervention and ample social support, families of children with Goldenhar's syndrome may

improve child and parent adjustment and the chances for healthy social-emotional outcomes.

Remaining Questions and Research Needs

Terminology, Definitions, and Causes

- Goldenhar's syndrome remains a mystery in many aspects. The many labels associated with the same set of anomalies causes confusion and creates difficulties in diagnosis and treatment.
- Diagnostic criteria are needed to define Goldenhar's syndrome and either separate the disorder from oculoauriculovertebral spectrum and hemifacial microsomia or unify these three conditions under one term.
- Although Tasse and colleagues (2005) have proposed a severity scoring system, this type of classification does not appear to be used in the mainstream. This system or a similar objective categorization needs to be implemented in order to further define Goldenhar's syndrome.
- Without this exactness of definition, the management of Goldenhar's syndrome may remain subjective and families may lack clarity in their understanding of the disorder.
- Further research must also be performed to ascertain the causes of Goldenhar's syndrome.
- Although some cases appear to be hereditary, no gene has been identified in the transmission of the disorder.

- The identification of such a gene would provide great knowledge about the syndrome and assist in genetic counseling and diagnosis.
- The link that appears to exist between Goldenhar's syndrome and assisted reproductive technology (ART) should be further investigated in order to identify the risks of ART and improve counseling for potential parents during this decision-making process.
- Similarly, more research is needed to further explore the relationship between Goldenhar's syndrome and maternal diabetes.
- The discrepancy in estimates of the incidence of Goldenhar's syndrome may be due in part to the lack of diagnostic criteria and the unspecified causes of the disorder.
- Incidence figures are necessary in order to understand ramifications of the syndrome in the general population and to prioritize screening and treatment procedures.
- Researchers should seek out better estimates of incidence and investigate any relationships between incidence and ethnicity or other birth factors.

Non-Medical Characteristics and Interventions

- While the medical research on Goldenhar's syndrome is rather extensive, gaps remain in the literature focusing on non-medical characteristics and interventions for the disorder.
- Due to the common characteristics of hearing loss and craniofacial anomalies, research is necessary in the areas of audiological assessment

and intervention, speech and language assessment and intervention, auditory skill development, and speech and language development.

- Although Goldenhar's syndrome remains relatively uncommon, practitioners must be prepared to provide treatment to these children, and they must have a solid research base to support their professional assessments and decisions.

Conclusion

- This webpage has reviewed the physical and behavioral characteristics of Goldenhar's syndrome and examined current practices in the management of children with this disorder. many children with Goldenhar's syndrome, medical and behavioral interventions become a way of life.
- Although Goldenhar's syndrome remains elusive on several levels, medical knowledge about the syndrome has led to relatively established procedures in medical intervention.
- However, rehabilitative and therapeutic interventions are far less standardized.
- Professionals in these areas must rely on research performed in related disorders in order to tailor treatment for individual children with Goldenhar's syndrome.
- Children with Goldenhar's syndrome comprise a small but important cross-section of children with congenital hearing loss. By performing further research into the causes and outcomes of Goldenhar's syndrome, practitioners may learn about embryological development, genetics, health

risks during pregnancy, and intervention strategies for children with craniofacial abnormalities. Further research is clearly needed in these areas.

- However, with current knowledge and good evidence-based practice, children with Goldenhar's syndrome and their families may receive the treatment and support needed to produce healthy and accomplished members of society.

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