Case study

Hearing loss and language delay in a child with Goldenhar syndrome: case report and literature review.

Abstract:

Goldenhar syndrome is a rare congenital disorder that involves the first and second branchial arches. It manifests mainly with asymmetric incomplete facial development, ear malformations, epibulbar dermoids and/or coloboma, and vertebral anomalies. It is constituted with a wide spectrum of signs and symptoms. Systemic anomalies may be associated. The etiology is still unclear. Ear malformations and hearing loss are very common. Early identification of auricular abnormalities is crucial in order to prevent secondary language and cognitive developmental delay. The purpose of this case report was to describe clinical presentation of Goldenhar syndrome in a 4-year-old female child who presented with language delay, and to discuss diagnosis and treatment of ear abnormalities and hearing loss.

Keywords: Goldenhar syndrome, ear malformations, hearing loss.

Introduction:

Goldenhar syndrome is a congenital disorder of craniofacial morphogenesis. It was originally identified by an ophthalmologist Maurice Goldenhar in a patient with a triad of mandibular hypoplasia, accessory tragi, and ocular dermoids in 1952. in 1963, Gorlin et al added vertebral anomalies and called it facio-auriculo-vertebral syndrome. (1) It is a group of congenital deformities due to a defect in the first and second branchial arches, which give

rise to the ear, face and eyelids. (2-3) Thus, patients present with unilateral asymmetric incomplete facial development, which may affect the maxillary bone, the mandible, ears, soft tissues and nerves, along with vertebral anomalies. (4-5) the etiology is not entirely understood, but it has been hypothesized that the defective formation of the branchial arches may be due to an abnormal embryonic vascular supply or disruption to the mesoderm. (6).

The prevalence of Goldenhar syndrome ranges from 1:3500 to 1:5600 live births, with a male to female ratio of 3:2, it is more common among infants with congenital deafness, occurring at a rate of about 1:1000 live births (7-8). Even though the majority of cases are sporadic, familial histories have been observed, suggesting autosomal or recessive inheritance. Some researchers suggested a multifactorial mode of inheritance where multiple genes interact, possibly in combination with environmental factors. Many risk factors have been proposed such as the use of thalidomide, tamoxifen, retinoid acid and cocaine during pregnancy, gestational diabetes, rubella and influenza. (9)

In this article, we report the case of a 4-year-old child with Goldenhar syndrome, with discussion on clinical features, especially auricular abnormalities and associated hearing loss.

Case report:

A 4-year-old orphan female child, with no medical past history, presented to our department with the complaint of language delay. We have no information about the child prenatal and birth circumstances or family medical history.

On clinical examination, there was facial asymmetry with hypoplasia of the left malar region, hypoplasia of the maxillary bone and the mandible, and a slight extension of

the labial commissure to the left. There was no facial palsy. Otoscopic examination showed grade 1 left microtia with preauricular tags and stenosis of the left external auditory canal. Ophthalmologic examination found left limbal dermoid cyst along with a lower eyelid coloboma. Hypertelorism was present. Intraoral examination didn't show any oral lesion. the palate and the tongue, the buccal and labial mucosae appeared to be normal. Cranial nerve examination didn't reveal any abnormality. General examination didn't find any systemic disease. We diagnosed the child as a case of Goldenhar syndrome. Hearing was assessed using auditory evoked potentials, the child was found to have a left hearing loss with a hearing threshold at 40dB. Hearing was normal in the right ear. The patient was advised to use hearing aids and participate in speech therapy.

Figure 1: photograph showing the frontal view of the patients face

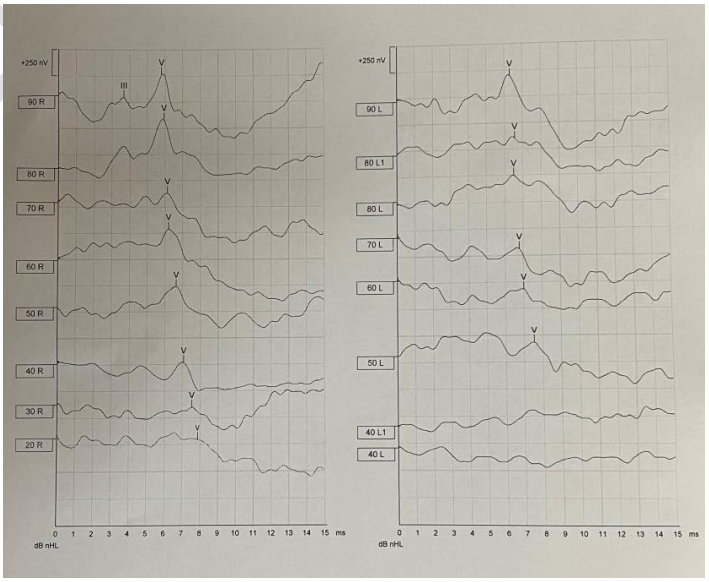

Figure 2: Auditory evoked potentials of the patient.

Discussion:

Goldenhar syndrome is an extremely rare condition. Symptoms and physical signs may vary greatly in range and severity from case to case. There are no specific criteria for diagnosis. Goldenhar syndrome is considered when two or more of the following anomalies are present: Hemifacial microsomia (including micrognathia), ear malformations (including microtia and accessory tragi), epibulbar dermoids and/or coloboma, and vertebral anomalies (fused or cervical hemivertebrae) (10). It is unilateral in 85% of cases, with involvement of right side to left side in a ratio of 3:2. Our patient presented with auricular abnormalities associated with mandibular hypoplasia, which is consistent with diagnostic criteria of Goldenhar syndrome. Other abnormalities may be associated such as ophthalmic malformations, cardiovascular, central nervous system, and genitourinary malformations, and may sometimes worsen the prognosis. Mental retardation is found in 6.5% to 15% of cases. (11). In the series of Rollnick et al (12), 294 patients with Goldenhar syndrome were included, 22% of patients had cleft lip or palate, 5% had cardiac anomalies, and 34% had skeletal anomalies. However, in 52% of cases, there were no associated anomalies. In the cohort of Morrison et al (13), 8 patients of 25 had cardiac anomalies. Furtado reported a tracheal stenosis in one patient. Digillio et al reported 2 patients with laryngomalacia. Our patient didn't have any of these associated abnormalities. (14)

Auricular abnormalities may affect the external, middle, or the inner ear. External and middle ear are more commonly affected (90%) than the inner ear (70%). (12) A recent systematic review included 62 records with 5122 patients. 52-100% of patients had ear anomalies. Microtia, pre-auricular tags, and atresia of the external auditory canal are the most reported external ear malformations. The most common middle ear malformations were ossicular anomalies, while the most reported inner ear abnormalities were oval window anomalies, cochlear anomalies, and anomalies of the semicircular canals. (15)

Hearing loss in patients with GS is rarely assessed in the cases that have been documented in the literature. In the review of Rooijers et al, hearing loss was reported in 29 to 100% of patients, which included conductive hearing loss (11.1 to 97%), mixed hearing loss (4.5 to 44.4%), and sensorineural hearing loss (1 to 40%).

Early detection of ear and hearing anomalies is crucial, as timely diagnosis enables early intervention, which subsequently prevents secondary developmental disorders of speech and language, as well as cognitive development. Patients with evident external ear malformations should be tested for hearing loss using age-appropriate audiology. Imaging tests might be indicated to evaluate associated middle or inner ear malformations and for pre-therapeutic assessment. (16) Surgical treatment such as placement of bone conduction devices may improve hearing at speech threshold. There is little information on the use of cochlear implant in the literature. (17) Since patients with no apparent external ear malformations may also suffer from hearing loss, it is recommended to evaluate audition in all patients with GS. The European guideline recommend that all new born with GS should undergo a neonatal hearing test. (18) If hearing loss is suspected, the infant should be referred to a specialized center for further evaluation. It is recommended to reassess hearing at the age of 24-30 months in all patients. audiological interventions should start as early as possible in order to prevent development delays. (18)

Treatment depends on the type of ear abnormality and severity of hearing loss. Given the heterogeneity of auricular anomalies, a case-by-case approach is necessary. A wide range of surgical procedures could be used, such as cochlear implantation, anterior tympanotomy, ossiculoplasty, and others. Surgical treatment usually consists on the correction of ear malformations and exision of preauricular tags. Hearing aids may be beneficial in patients with minor external malformations. The Bone Anchored Hearing Aid (BAHA) is an excellent option for severe bilateral microtia with atresia and conductive hearing loss. (15)

Conclusion:

Ear malformations and hearing loss are common in patients with Goldenhar syndrome. Early detection and intervention prevent secondary developmental language and cognitive disorders.

Ethical approval:

As per international standard or university standard written ethical approval has been collected and preserved by the author(s).

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