Audiological Findings in Patients with Oculo-Auriculo-Vertebral Spectrum

Pricila Sleifer[1] Natalya de Souza Gorsky[2] Thayse Bienert Goetze[3] Rafael Fabiano Machado Rosa[4] Paulo Ricardo Gazzola Zen[5]

[1] Department of Health and Human Communication, Universidade Federal do Rio Grande do Sul, Porto Alegre, Rio Grande do Sul, Brazil
[2] Curso de Fonoaudiologia, Universidade Federal do Rio Grande do Sul, Porto Alegre, Rio Grande do Sul, Brazil
[3] Mestranda Programa de Pós-Graduação em Patologia, Universidade Federal de Ciências da Saúde de Porto Alegre, Porto Alegre, Rio Grande do Sul, Brazil
[4] Professor Colaborador do Programa de Pós-Graduação em Patologia, Universidade Federal de Ciências da Saúde de Porto Alegre, Porto Alegre, Rio Grande do Sul, Brazil
[5] Professor do Programa de Pós-Graduação em Patologia, Universidade Federal de Ciências da Saúde de Porto Alegre, Porto Alegre, Rio Grande do Sul, Brazil

Address for correspondence Pricila Sleifer, PhD, Professor, Department of Health and Human Communication, Universidade Federal do Rio Grande do Sul, Rua Ramiro Barcelos, 2600 Instituto de Psicologia Porto Alegre, Rio Grande do Sul 90040060, Brazil (e-mail: pricilasleifer@uol.com.br).

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Abstract

Introduction Oculo-auriculo-vertebral spectrum, also referred to as *Goldenhar syndrome*, is a condition characterized by alterations involving the development of the structures of the first and second branchial arches. The abnormalities primarily affect the face, the eyes, the spine, and the ears, and the auricular abnormalities are associated with possible hearing loss.

Objective To analyze the audiological findings of patients with oculo-auriculo-vertebral spectrum through liminal pure-tone audiometry and speech audiometry test.

Methods Cross-sectional study conducted on 10 patients with oculo-auriculo-vertebral spectrum and clinical findings on at least two of the following areas: orocraniofacial, ocular, auricular, and vertebral. All patients underwent tonal and vocal hearing evaluations.

Results Seven patients were male and three were female; all had ear abnormalities, and the right side was the most often affected. Conductive hearing loss was the most common (found in 10 ears), followed by sensorineural hearing loss (in five ears), with mixed hearing loss in only one ear. The impairment of the hearing loss ranged from mild to moderate, with one case of profound loss.

Conclusions The results show a higher frequency of conductive hearing loss among individuals with the oculo-auriculo-vertebral spectrum phenotype, especially moderate loss affecting the right side. Furthermore, research in auditory thresholds in the oculo-auriculo-vertebral spectrum is important in speech therapy findings about the disease to facilitate early intervention for possible alterations.

Keywords
- Goldenhar syndrome
- hearing
- speech
- language and hearing science

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Introduction

The oculo-auricular-vertebral spectrum (OAVS) was first described by Maurice Goldenhar in 1952[1], which is why it is also known as *Goldenhar syndrome*, or hemifacial microsomia. It is a rare condition,[2] characterized by alterations involving mainly the face, eyes, ears, and spine.[3] OAVS is related to a blastogenesis dysfunction, which affects the first and second pharyngeal arches.[4] Some of the identified OAVS characteristics are hemifacial microsomia, mandibular hypoplasia, epibulbar dermoids, and skeletal anomalies.[1] Moreover, the right side is more often affected than the left.[5]

This condition's etiology is not yet known, but arguably it is associated with environmental and nutritional factors, such as maternal drug ingestion or diabetes.[6] OAVS incidence varies from 1:5,200 to 1:26,500 live births and affects boys more often than girls.[7]

Of all OAVS clinical manifestations, this article will focus on auricular abnormalities and how they affect the auditory system. The external, middle, and inner ear can be affected in patients with OAVS, but abnormalities prevail on the external and middle ear (90%) rather than the inner ear (70%).[8] The most common external ear abnormalities are microtia and external auditory canal atresia. In the middle ear, it is common to find anomalies in the ossicular chain and otitis media with effusion. Despite not widely referenced in literature, inner ear abnormalities can also occur, affecting the cochlea and semicircular canals.[9]

There is a shortage of published studies in the national literature that specifically detail findings related to auricular, and consequently, auditory abnormalities due to OAVS. A search through the bibliography available on SciELO and PubMed databases, using as keywords "oculo-auriculo-vertebral spectrum," "Goldenhar syndrome," and "hearing," found few works between 2001 and 2013 pertaining exclusively to OAVS and auricular abnormalities.

Considering the clinical relevance of the subject and the need for more contributions to expand general data on OAVS, this article aims to analyze the auditory findings of patients affected by OAVS through liminal pure tone and speech audiometry tests.

Methods

This contemporary cross-sectional study examined the analysis of auditory findings on patients affected by OAVS. This study was approved by the research ethics committee under number 851/09 on May 14, 2009. All patients, parents, and/or legal guardians signed a free and informed consent form before the beginning of data collection and evaluation.

Ten patients with OAVS were assessed and presented clinical abnormalities in at least two of the following areas: orocraniofacial, ocular, auricular, and vertebral. This approach was the same adopted by Strömland et al.[10] All selected patients were diagnosed with OAVS at the Universidade Federal de Ciências da Saúde de Porto Alegre Clinical Genetics Service. Auricular abnormalities were described according to the affected side and segment, divided into external, middle, and inner ear.

Patients selected for this study were evaluated at hearing electrophysiology studies of Universidade Federal do Rio Grande do Sul. They initially underwent a specific anamnesis, received instructions about the tests, and finally underwent pure tone and speech audiometry tests. Evaluations were performed in sound booths with AC40 and AD229 interacoustics audiometers and TDH39 headphones.

The first test was the liminal pure tone audiometry of the air conduct at 250, 500, 1,000, 2,000, 3,000, 4,000, 6,000, and 8,000 Hz. The frequencies of 500, 1,000, 2,000, 3,000, and 4,000 Hz were tested on the bone conduct. Stimuli were introduced using the gradient descent method. Results were analyzed by calculating the average of 500-, 1,000-, and 2,000-dB frequencies. The degrees of hearing loss were classified according to Davis and Silverman's criteria.[11]

The speech audiometry test was started by observing the speech recognition threshold (SRT), during which audible three-syllable words were introduced to the patient, 40 dBHL above the tritonal average in the air conduct. This intensity was reduced until the threshold was reached. Each patient was instructed to repeat the words heard, and the SRT was considered at the intensity that allowed patients to correctly reproduce 50% of the words heard.

After that, the percentage index of speech recognition (PISR) was measured. The PISR consisted of introducing a list of 25 audible monosyllable words to the patients, 40 dBHL above the tritonal average in the air conduct. Each patient was instructed to repeat the words heard. If a subject answered correctly 92 to 100% of times, the patient was considered to have no anomalies; if a patient responded correctly less than 88% of the time, another 25 two-syllable words were introduced and a new percentage of reproduction was recorded. When a patient was not able to perform the SRT and the PISR, the speech detection threshold (SDT) was applied.[12] Some patients needed to conduct reviews in more than 1 day of attendance.

Data were analyzed with the Statistical Package for Social Sciences software, version 17.0 (SPSS Inc., Chicago, Illinois, United States) for Windows, adopting a significance level of 5% for statistic decisions criteria. The McNemar test was used to compare right and left ears.

Results

The sample was composed of 10 patients, with an average age of 10.1 ($\pm$6.6) years and a minimum of 3 and maximum of 27 years (median of 9 years). Male patients predominated in the sample (70%; ►**Table 1**).

We observed a predominance of abnormalities on the external ear on both left and right sides, each representing 50% ($n = 5$). Auricular abnormalities found included microtia, preauricular appendages, ear lobe and external auditory canal agenesis, anotia, malformed ossicular chains, malformed ossicles, and reduced oval window. One patient also had a cleft lip and palate.

Table 1 Gender, age, and diagnosis distribution and measures of central tendency and variability for age

Variables	Total sample ($n = 10$)	
	n	%
Gender		
Female	3	30.0
Male	7	70.0
Age (y)		
Mean ± standard deviation	10.1 ± 6.6	
Median (range)	9.0 (3–27)	
Auricular abnormalities LE		
External ear	5	50.0
Middle ear	1	10.0
Inner ear		
No change	2	20.0
External and middle ear	1	10.0
Middle and inner ear	1	10.0
Auricular abnormalities RE		
External ear	5	50.0
Middle ear		
Inner ear		
No change	2	20.0
External and middle ear	2	20.0
Middle and inner ear	1	10.0
Type of loss LE		
Normal auditory thresholds	3	30.0
Conductive hearing loss	4	40.0
Sensorineural hearing loss	2	20.0
Loss mixed hearing	1	10.0
Type of loss RE		
Normal auditory thresholds	1	10.0
Conductive hearing loss	6	60.0
Sensorineural hearing loss	3	30.0
Loss mixed hearing		
Degree of loss LE		
Normal auditory thresholds	3	30.0
Mild	4	40.0
Moderate	2	20.0
Profound	1	10.0
Degree of loss RE		
Normal auditory thresholds	1	10.0
Mild	3	30.0
Moderate	5	50.0
Profound	1	10.0

Abbreviations: LE, left ear; RE, right ear.

The auricular abnormalities observed mainly affected the external ear on the left (70%) and right side (80%). The left ear displayed more conductive hearing loss (40%). This also happened with the right ear (60%). The left ear showed more occurrences of mild loss (40%), whereas the right ear had more occurrences of moderate loss (50%; ►**Table 2**).

However, in statistic analysis the McNemar test showed no significant difference between ears in type or degree of hearing loss (►**Table 2**).

Discussion

Results showed that males were more commonly affected in this study, constituting 7 out of the 10 patients of the sample. This finding is in agreement with other studies in which the male sex was also the most affected.[7,9,13,14] One study about OAVS and auricular abnormalities related the female gender as more affected, as opposed to the present study.[15]

Auricular abnormalities found on selected patients affected mainly the external ear, 70% on the left side and 80% on the

Table 2 Headset, type, and degree of hearing loss change distribution according to ear

Variables	Ear ($n = 20$)				p[a]
	LE		RE		
	n	%	*n*	%	
Auricular abnormalities					
External ear	5	50.0	5	50.0	
Middle ear	1	10.0			
Inner ear					> 0.999
No change	2	20.0	2	20.0	
External and middle ear	1	10.0	2	20.0	
Middle and inner ear	1	10.0	1	10.0	
Type of loss					
Normal auditory thresholds	3	30.0	1	10.0	
Conductive hearing loss	4	40.0	6	60.0	0.702
Sensorineural hearing loss	2	20.0	3	30.0	
Loss mixed hearing	1	10.0			
Degree of loss					
Normal auditory thresholds	3	30.0	1	10.0	
Mild	4	40.0	3	30.0	0.552
Moderate	2	20.0	5	50.0	
profound	1	10,0	1	10,0	

Abbreviations: LE, left ear; RE, right ear.
[a]McNemar test.

right. These findings are consistent with those described in the literature reviewed.[1,6,8,9,14,15]

Little information is available about the type of hearing loss detected in patients with OAVS in other studies. In addition, in almost all cases, the degree of hearing loss is not specified. This information should be further studied and reported so that OAVS auricular abnormalities can be better evaluated and related to the type and degree of possible hearing loss.

We verified that right ears were more often affected than left ears. Other studies also indicated this finding.[1,5,16]

Conductive hearing loss was the most frequent type observed in this study, occurring in 10 ears. It ranged from mild to moderate loss. A study about auricular abnormalities in nine OAVS patients described anomalies on the external and middle ear in 12 and 9 ears, respectively.[9] This type of anomaly was considered by the authors as an indication of possible conductive hearing loss, as it is directly related to the components deemed abnormal in these patients.

This factor is also associated with the embryology of external and middle ears, which develop after the first and second pharyngeal arches. OAVS is directly related to a blastogenesis dysfunction involving these two arches, which explains the auricular abnormalities found on patients diagnosed with this condition.[17]

Sensorineural hearing loss was also found in five ears of the selected patients, of mild ($n = 2$), moderate ($n = 1$), and profound ($n = 2$) degrees. This type of hearing loss was also diagnosed in patients with OAVS in other studies.[1,5,6,15,18] The presence of sensorineural hearing loss can indicate that other components of the embryonic formation of individuals with OAVS might be affected, beyond the first and second pharyngeal arches.

A study searching for inner ear anomalies in a group of five patients with OAVS observed conductive hearing loss in three of them.[14] In addition to inner ear abnormalities, patients also had middle and external ear anomalies. Similarly, another study identified auricular abnormalities on the inner ear in nine patients.[9]

The inner ear abnormalities and consequent sensorineural hearing loss could be associated with the migration of neural crest cells during the embryonic period, which might indicate more components involved in the development of patients with OAVS.[5,14,17]

There was also a case of mixed hearing loss in one ear in the sample used for this study. This type of hearing loss was the most frequently observed in a different study.[18] The mixed hearing loss demonstrated the involvement of other embryonic structures beyond the first and second pharyngeal arches. In addition, four ears showed normal auditory thresholds.

Moderate loss was the most frequently observed, present in seven ears. Mild hearing loss was verified in six ears. Profound hearing loss was identified in both ears of one patient. As conductive hearing loss occurred more often, mild and moderate degrees were observed, because the anatomy and physiology of conductive hearing losses does not present an air–bone gap superior to 60 dB, meaning it hardly achieves profound degrees. Profound loss has been associated with sensorineural hearing loss. No case showed a severe degree of hearing loss.

SRT results in all patients were compatible with tritonal averages of frequencies of 500, 1,000, and 2,000 Hz used in pure tone audiometry. The PISR results were equal or superior to 92% in the conductive hearing losses and inferior to 88% in the sensorineural or mixed hearing losses. In one case of profound sensorineural hearing loss, it was necessary to apply the SDT for both ears.

Although auricular malformations and hearing loss were more often observed in the right ear, the statistic analysis performed with the McNemar test did not show any significant difference between ears and type of hearing loss. Furthermore, there were also no significant differences between ears and degree of hearing loss. Perhaps there could be a statistically significant difference in a sample with more individuals.

When the hearing loss and auricular malformation were compared, in the majority of findings, malformations affected the external and middle ear and they caused conductive hearing loss. Nonetheless, the presence of sensorineural hearing losses was also found without single commitment of inner ear and in one case of compromised external ear and profound sensorineural hearing loss. These last findings suggest the use of diagnostic imaging for detecting possible anomalies associated with OAVS.

In one case study, a female patient was diagnosed with OAVS. The patient showed microtia and high-set right ear.[4] The patient's auditory evaluation revealed a moderate mixed hearing loss on the right ear and mild sensorineural hearing loss on the left ear. In addition to that, she also presented speech anomalies such as phonological disorders.

Another study conducted with a group of 11 patients with OAVS explored possible surgical interventions to correct auricular abnormalities and consequent hearing loss.[8] Two patients with profound sensorineural hearing loss were selected to undergo cochlear implant surgery. After the implant placement, improvements were seen in the response to stimuli, varying from 100 dB to 50 dB in one of the patients and response to auditory stimuli at 40 dB in the other. The present study also observed profound sensorineural hearing loss in one of the evaluated patients. The adequate response of the aforementioned patients shows that it is possible to use a cochlear implant with patients with OAVS and obtain satisfactory results.

Another study reported on a case study of a child with OAVS showing moderate conductive hearing loss on the right ear and progressive hearing loss on the left ear.[16] This study implemented the use of a frequency modulation system in the classroom. The system was used on the left ear and resulted in satisfactory improvements of the child's school competence.

These findings show that hearing losses associated with OAVS may negatively affect a child's speech and interfere in other aspects such as their school life. Adequate diagnosis of hearing dysfunctions associated with OAVS may help in the search and implementation of early auditory interventions as well as speech and language therapies, which ensures better quality of life to the patients.

Conclusion

Conductive hearing loss is often observed in individuals with OAVS. It is usually of moderate degree and more often affects the right side. This is arguably related to the auricular abnormalities found and is common in patients with OAVS, which affects the conductive component of the auditory system.

Moreover, the study of auditory thresholds in patients with OAVS is important to further the phonoaudiological findings about the condition, as it aids in diagnosis and enables early intervention for the possible abnormalities found.

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