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Ophthalmic pathologies in female subjects with bilateral congenital sensorineural hearing loss

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Background/aim: The high prevalence of ophthalmologic pathologies in hearing-disabled subjects necessitates early screening of other sensory deficits, especially visual function. The aim of this study is to determine the frequency and clinical characteristics of ophthalmic pathologies in patients with congenital bilateral sensorineural hearing loss (SNHL).

Materials and methods: This descriptive study is a prospective analysis of 78 young female SNHL subjects who were examined at a tertiary care university hospital with a detailed ophthalmic examination, including electroretinography (ERG) and visual field tests as needed.

Results: The mean age was 19.00 ± 1.69 years (range: 15 to 24 years). A total of 39 cases (50%) had at least one ocular pathology. Refractive errors were the leading problem, found in 35 patients (44.9%). Anterior segment examination revealed heterochromia iridis or Waardenburg syndrome in 2 cases (2.56%). Dilated fundus examination revealed retinal pathologies in 15 cases (19.23%), including retinitis pigmentosa or Usher's syndrome in 8 cases (10.25%). Most of the Usher's syndrome cases (87.5%) had consanguinity.

Conclusion: Screening for congenital SNHL in the early years of life and routine yearly follow-ups are essential for maximizing the rehabilitation of this disabled group. The high rate of visually debilitating syndromic ocular pathologies associated with high frequency of consanguinity doubles the importance.

Key words: Consanguinity, ophthalmologic screening, retinitis pigmentosa, sensorineural deafness, Usher's syndrome

1. Introduction

Sensorineural hearing loss (SNHL) is a challenging condition with profound social, cultural, and medical effects for patients, families, and society (1). The prevalence of bilateral, moderate-to-severe congenital SNHL is 1–3 per 1000 live births worldwide (2,3). A congenitally deaf individual may have developmental delays in language, speech, and cognitive and social development (4,5). Due to the absence of auditory input, they are more dependent on their vision in order to compensate. Like hearing, vision plays an important role in gathering information from the environment. Visual and auditory channels are responsible for more than 95% of environmental cognition (6,7), so early identification and correction of any ophthalmic problems are essential (8,9).

The prevalence of ophthalmologic pathologies in hearing-impaired and deaf children has been reported to be as high as 45.8% (10). For this reason, early screening for other sensory deficits, especially visual function, and immediate treatment are essential (4). In addition, the ophthalmologic examination may also help to diagnose syndromic causes of hearing loss like Usher's and Waardenburg syndromes (11). Hearing and visually impaired patients are significantly disabled, being less able to lip read, less cooperative, and less capable of manual tasks than hearing-impaired children with normal vision (12). These syndromic cases are especially preferred candidates for early cochlear implants, which will improve listening and spoken language skills (13).

In this study, we aimed to investigate the burden of visual disability among young women with bilateral congenital SNHL.

2. Materials and methods

This descriptive study is an analysis of 78 SNHL cases from Kemal Yurtbilir Hearing Impaired Vocational

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High School for hearing impaired students in Turkey. The study was performed at Gülhane Medical Academy, in adherence to the guidelines of the Declaration of Helsinki, and was approved by the ethics committee of Gülhane Medical Academy (05.03.2013/No:14).

The participants were 78 young females aged 19.00 ± 1.69 years (range: 15 to 24 years). All had bilateral SNHL above 70 dB confirmed by established laboratory tests. None of patients had previously received a cochlear implant. All patients' hearing loss was prelingual (before 18 months). None of patients had systemic disease except for hearing loss.

The participants were evaluated with a detailed ophthalmic examination, with the help of a trained sign language teacher and a nurse near them.

Detailed ophthalmic examination included, in order of administration, refraction, best corrected visual acuity (BCVA), stereoacuity, ocular motility, slit lamp examination, and dilated fundus examination with 0.5% tropicamide. Patients with indications of tapetoretinal dystrophy underwent electroretinography (ERG) and visual field tests.

Refraction was evaluated with an auto kerato-refractometer (Topcon KR-8800). The refractive errors were considered to be clinically significant when hypermetropia, myopia, or astigmatism were greater than or equal to ±1.0 diopters. Anisometropia was defined as a difference of ≥1.0 diopters between the eyes (10).

BCVA was evaluated with the Snellen visual acuity chart and was translated into the equivalent logarithm of the minimal angle of resolution (LogMAR).

Amblyopia was defined as ≥2 lines BCVA difference between the eyes resulting from refractive, strabismic, or deprivational sources in the absence of an organic pathology.

Stereoacuity was evaluated with the Randot test, and ≥100 s of arc was considered to be reduced depth perception.

Ocular motility was evaluated with Hirschberg and cover–uncover tests. Strabismus was diagnosed when misalignment exceeded 10 prism diopters.

ERG (Roland-Consult) was performed and evaluated according to the International Society for Clinical Electrophysiology of Vision (ISCEV) standards (14).

Visual field tests were performed using automated static perimetry (Humphrey Field Analyzer, Model 750i, Carl Zeiss Meditec, Inc., Dublin, CA, USA). Low patient reliability was defined as fixation loss over 20% and false-positive and false-negative results over 15%. The SITA 30-2° and 60-4 fast program was used.

Statistical analyses were performed using SPSS 16 (SPSS, Chicago, IL, USA). For descriptive statistics, discontinuous variables were shown as numbers and percentage (%); continuous variables were shown as mean ± standard deviation and median (minimum–maximum).

3. Results

A total of 78 cases of SNHL were analyzed in this study. All of the patients were young women. The mean age was 19.00 ± 1.69 years (range: 15 to 24 years).

A total of 39 cases (50%) had at least one ocular abnormality. Refractive errors were the leading pathology, found in 35 cases (44.9%), while 22 cases (28.2%) had more than one ocular problem (Tables 1 and 2).

Table 1. Refractive errors and visual acuity assessments of the subjects.

Pathology	Number of cases	Percentage (%)
Refractive errors		
Emmetropia	43	55.1
Myopia	20	25.7
Hyperopia	7	8.9
Myopic Astigmatism	6	7.8
Hyperopic Astigmatism	2	2.6
BCVA (LogMAR)		
0.0	67	85.8
0.1	5	6.4
0.2	4	5.2
0.5	1	1.3
0.7	1	1.3

BCVA = best corrected visual acuity; LogMAR = logarithm of the minimal angle of resolution.

Table 2. Summary of ocular pathologies in patients with bilateral congenital SNHL.

Pathology	Number of cases	Percentage (%)
Amblyopia	4	5.12
Reduced stereopsis	9	11.5
Strabismus	4	5.12
Esotropia	1	1.28
Exotropia	1	1.28
Hypotropia	1	1.28
Duane syndrome	1	1.28
Ptosis	1	1.28
Heterochromia iridis/Waardenburg syndrome	2	2.56
Retinal pathologies	15	19.2
Usher syndrome	8	10.25
RPE disturbances	5	6.41
Myelinated nerve fibers	2	2.56

SNHL = sensorineural hearing loss; RPE = retinal pigment epithelium.

BCVA was less than 0.0 LogMAR in 11 cases (14.1%). Of these, in 5 cases (6.41%) BCVA was 0.1 LogMAR, in 4 cases (5.12%) BCVA was 0.2 LogMAR, in 1 case (1.28%) BCVA was 0.5 LogMAR, and in 1 case (1.28%) BCVA was 0.7 LogMAR (Table 1).

Amblyopia was detected in 4 cases (5.12%), all of which were caused by anisometropia. Reduced depth perception was found in 9 cases (11.53%). Anterior segment examination revealed heterochromia iridum in 2 cases (2.56%); those patients were diagnosed with Waardenburg syndrome. Four patients (5.12%) had strabismus, including 1 case (1.28%) with esotropia, 1 case (1.28%) with exotropia, 1 case (1.28%) with hypotropia, and 1 case (1.28%) with bilateral Duane syndrome type 1. External ocular examination revealed ptosis in 1 case (1.28%). The summary of ocular pathologies is listed in Table 2.

Dilated fundus examination findings suggestive of tapetoretinal dystrophy were present in 8 cases (10.25%). All of these patients underwent ERG. All test results were abnormal, and the patients were diagnosed with retinitis pigmentosa (RP). Due to the coexistence of RP and bilateral congenital SNHL, all 8 patients (10.25%) were also diagnosed with Usher's syndrome. Clinical characteristics of the Usher's syndrome cases are listed in Table 3. RP was classified as early stage in 3 cases (37.5%), middle stage in 1 case (12.5%), and end stage in 4 cases (50%) according to history, clinical examination, ERG, and 30-2 visual field tests (15). Additional ophthalmic pathologies in the Usher's syndrome cases were posterior subcapsular cataract in 3 cases (37.5%) and cystoid macular edema (CME) in 1 case (12.5%). Family history of Usher's syndrome cases revealed 87.5% consanguinity with first-degree relatives.

Other retinal pathologies were retinal pigment epithelium (RPE) disturbances in 5 cases (6.41%) and myelinated nerve fibers in 2 cases (2.56%) (Table 2).

4. Discussion

Published studies have shown that the rate of ophthalmologic pathologies in SNHL cases ranges between 31% and 48% (4,7–11,16–19). A summary of frequency of ophthalmic pathologies from different studies is listed in Table 4. The variability of ophthalmic pathologies across different studies is likely secondary to the differences in patient age range, sample size, examination techniques, and the criteria used to define visual impairment.

This study showed a slightly higher incidence (50%) of ocular pathologies in congenitally deaf patients. In the same manner, refractive disorders were the leading pathology, with a higher incidence of 44.9% (35 cases), compared to the 16%–42.7% found across previously published studies (4,7,9–11,16–20). Since our study group was composed of adult individuals, it was easy to detect even a small degree of refractive problems like myopia, which may increase during the adolescent years.

Manifest strabismus was detected in 5.12% of our sample (4 cases), which was consistent with most previous studies (4,7,9,11,17,20), although some found higher results of up to 18.2% (16) and 24.1% (10).

Anterior segment examination revealed heterochromia iridum and Waardenburg syndrome in 2.56% of our sample (2 cases), which was reported as between 0% and 3.6% in previous studies (4,7,9–11,13,16–20). A summary of frequency of Usher's syndrome and Waardenburg syndrome from different studies is listed in Table 4.

Table 3. Clinical features of the Usher syndrome cases.

Clinical features	Case 1	Case 2	Case 3	Case 4	Case 5	Case 6	Case 7	Case 8
Nyctalopia	Severe	Mild	Moderate	Mild	Mild	Severe	Moderate	Mild
BCVA right/left (LogMAR)	0.1/0.2	0.0/0.0	0.1/0.1	0.1/0.2	0.1/0.1	0.7/0.7	0.0/0.1	0.1/0.1
Refraction	MA	MA	MA	MA	MA	MA	MA	MA
Pigmentation area	Mid-peripheral	Peripheral	Mid-peripheral	Peripheral	Peripheral	Mid-peripheral	Peripheral	Mid-peripheral
Retinal arteriolar narrowing	Prominent	Absent	Mild	Mild	Absent	Prominent	Absent	Prominent
Optic disc	Severely pallor	Normal	Mild pallor	Mild pallor	Normal	Severely pallor	Normal	Severely pallor
Additional pathologies	CME	-	PSCC	-	-	PSCC	-	PSCC
Visual field	Not reliable	Superior peripheral defect	Tunnel vision	Not reliable	Superior peripheral defect	Tunnel vision	Superior peripheral defect	Not reliable
ERG	Pathologic	Pathologic	Pathologic	Pathologic	Pathologic	Pathologic	Pathologic	Pathologic
Consanguinity	+	+	+	-	+	+	+	+
RP stage	End	Early	End	Mid	Early	End	Early	End

BCVA = best corrected visual acuity; LogMAR = logarithm of the minimal angle of resolution; MA = myopic astigmatism; CME = cystoid macular edema; PSCC = posterior subcapsular cataract; ERG = electroretinography; RP = retinitis pigmentosa.

Table 4. Literature data for ophthalmological findings in SNHL.

Year	Study group	Sample size	Age	Total ocular pathology	Refractive disorders	Strabismus	ERG	Usher syndrome	Waardenburg syndrome
1995	Armitage et al. (10)	83	0–10 y/o	45.8%	28.9%	24%	No	0%	3.6%
2000	Brinks et al. (17)	238	10–21 y/o	48%	16%	3%	Yes	2.2%	0%
2000	Mets et al. (13)	48	6 mo–9 y/o	Unknown	Unknown	Unknown	Yes	10.4%	0%
2002	Mafong et al. (4)	49	1–18 y/o	31%	18.4%	2.04%	No	0%	0%
2003	Hanioğlu et al. (16)	104	7–20 y/o	40%	29.8%	18.2%	Only if needed	0.9%	0.9%
2003	Guy et al. (9)	110	8 mo–16 y/o	43.6%	42. %	6.36%	78 of 110	5.45%	0%
2009	Bakhshaee et al. (7)	50	3–7 y/o	32%	28%	6%	No	4%	0%
2009	Sharma et al. (20)	226	0–18 y/o	21.7%	10.2%	3%	unknown	1.3%	0.9%
2010	Falzon et al. (19)	141	16 mo–9 y/o	41.8%	26.4%	8.4%	Only if needed	1.4%	0.7%
2011	Ceyhan et al. (11)	208	7–19 y/o	34.6%	29.8%	2.4%	No	5.8%	1.4%
2011	Bist et al. (18)	279	5–20 y/o	28%	16.48%	Unknown	Only if needed	0.36%	0.36%
2014	Our study	78	15–24 y/o	50%	44.9%	5.12%	Only if needed	10.25%	2.56%

SNHL = sensorineural hearing loss; y/o = years old; mo = months.

We found Usher's syndrome in 10.25% of our sample (8 cases). Although ERG was performed only if needed, there was a higher frequency of Usher's syndrome in our study than in previously published reports (4,7,9–11,13,16–20). Rubella retinopathy was not diagnosed in any cases in our study, although it has been previously reported at a rate of between 2.4% and 21% (7,16,17).

Some researchers (13) performed ERG as a screening test for all SNHL participants, and 10.4% of patients were diagnosed with Usher's syndrome. In another study (9), 78 of 110 participants completed ERG tests, and 5.45% of patients were diagnosed with Usher's syndrome. Although routine ERG testing was performed for all cases in another study (17), Usher's syndrome was diagnosed in only 2.2% of cases. As the authors described, they recruited the participants by letter; patients previously diagnosed with Usher's syndrome probably did not enter the study group. In one study (10) in which ERG was not performed, no cases of Usher's syndrome were diagnosed. In addition, that study group was in the first decade of life when RP is not clinically apparent, so the study may have underestimated the frequency of Usher's syndrome.

We think that we found a high frequency of Usher's syndrome in congenital bilateral SNHL cases for two major reasons. First, we examined an adult group. The clinical properties of RP were apparent in this age group, and we performed ERG tests with all patients who were suspected of having tapetoretinal dystrophy. Second, the high rate of consanguinity (87.5%) in the Usher's syndrome cases was a striking property of autosomal recessive diseases. Prevention of consanguinity through public health programs is therefore essential. Only Mets et al. (13) previously reported an association between Usher's syndrome and consanguinity.

Because of the high frequency of ophthalmic pathologies, we recommend detailed ophthalmic examination as soon as the diagnosis of permanent SNHL is confirmed and at any time if parents or educators suspect a problem. Yearly follow-ups and ERG tests as screening programs are essential in this regard. A yearly follow-up may detect changes in refractive errors and later-onset RP. Routine ERG tests for all bilateral SNHL cases should be performed as early in childhood as possible to screen for Usher's syndrome (4,9,13,17). Guy et al. (9) recommended screening all congenital SNHL patients with ERG after the age of 7. ERG has a key role in the diagnosis of Usher's syndrome and, if it is not performed, the frequency of Usher's syndrome may be underestimated. ERG is a noninvasive test that can identify RP before the onset of clinical findings (4). As retinal changes may be evident in the teenage years, follow-up ophthalmologic examinations of those patients for whom ERG has not been performed are essential (4). If there are any symptoms of Usher's syndrome such as night blindness, visual field loss, or unexplained reduction in visual acuity, ERG can be repeated in the following years. ERG is also a unique measure for differentiation of RP and other retinopathies, including rubella retinopathy (17).

For patients and parents who already have difficulties with a hearing disability, an additional progressive visual deterioration like Usher's syndrome may cause shock when no curative treatment can be offered. Some patients and parents may need more time to absorb the diagnosis. A follow-up appointment within 2 weeks should be offered to this group so that the parents and patient can discuss further questions.

In this regard, pediatricians, primary care physicians, special educators, ophthalmologists, and ear, nose, and throat specialists should be aware of the importance of ophthalmic screening and management of this special group.

There are several limitations of this study. First, we did not perform ERG with all 78 SNHL cases because this test is a demanding procedure. We only performed the test when Usher's syndrome was clinically suspected, and this was the weakest point of our study. Although we performed detailed examinations, the frequency of Usher's syndrome might be over the 10.25% that we identified. Second, all participants in the study were female because the boarding school only accepted female students.

A strength of this study was that the sample of SNHL patients included all boarding students of a special education high school, who were from different geographic areas of the country. In addition, the examinations and tests were performed by the same experienced examiners.

Early diagnosis of hereditary hearing loss syndromes like Usher's syndrome and Waardenburg syndrome is important for earlier cochlear implantation, language development, slowing of the progression of visual loss, genetic counseling, future therapies, and emotional regulation for patients and their families (4,9,21,22).

An unusual finding in this series was a high prevalence of ocular pathologies, especially Usher's syndrome — not only the proportion but also the frequency of consanguinity (87.5%). We conclude that congenital bilateral SNHL patients who already have an auditory sensory deficit have a greater dependence on visual input, so they should be given every opportunity to maximize their visual potential at the earliest age possible.

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