

# Importance of ophthalmological examination in children with congenital sensorineural hearing loss

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## ABSTRACT

**الأهداف:** تقييم أهمية فحص العيون في الأطفال المصابين بصمم عصبي خلقي (SNHL).

**الطريقة:** أجريت الدراسة في قسم الأنف و الإذن و الخنجرية – مستشفى الرمادي التعليمي –مدينة الرمادي – العراق خلال الفترة 20 ديسمبر 2007 حتى 30 أكتوبر 2008. اشتملت هذه الدراسة الاستطلاعية على 50 طفل مصاب بصمم عصبي. تم إجراء فحص العيون لجميع المرضى.

**النتائج:** وجدت اضطرابات في العيون لدى 16 (32%) مريض من 50 مريض مصابين بصمم عصبي خلقي. كانت أكثر إصابات العيون في الفئة العمرية بعد تعلم اللغة (81.3%) مقارنة بالمجموعات الأخرى و كانت ذي نتيجة إحصائية هامة ( $p=0.007$ ). قصر البصر هو أكثر اضطرابات العيون حيث بلغت نسبته 5 (31.3%) حالات، إصابته (12.5%) بعد البصر، إصابته بحول (12.5%)، إصابته بالتهاب الشبكية الصباغي و إصابة واحدة (6.3%) بالتهاب الجفن. كان لدى 4 مرضى آخرين عدة إصابات بالعين، الأول منهم مصاب بقصر البصر مع استجماتيزم (لابؤرية)، الثاني مصاب بعد البصر و التهاب المتحممة، الثالث التهاب جفني العينين مع حساسية العين، أما الأخير فكان مصابا بقصر البصر و التهاب جفن العين. بفحص العيون تم التوصل إلى تشخيص متلازمة فقدان السمع في طفلين (متلازمة أشر).

**خاتمة:** إصابات العيون من المشاكل الشائعة لدى المرضى المصابين بصمم عصبي خلقي، فمن الضروري فحص العيون لديهم لاكتشاف أي خلل في حدة الرؤية و تشخيص متلازمة فقدان السمع.

**Objectives:** To evaluate the importance of ophthalmological examination in children with congenital sensorineural hearing loss (SNHL).

**Methods:** This study was conducted at the Ear, Nose, and Throat Department of Al-Ramadi Teaching Hospital in Al-Ramadi city, Iraq from 20 December

2007 to 30 October 2008. Fifty children with congenital SNHL were included in this prospective study. Ophthalmological examination was carried out for all patients.

**Results:** Out of 50 patients with SNHL, 16 (32%) had ocular abnormalities. Ocular abnormalities were more common in postlingual age group (81.3%) than other age groups, which is statistically significant ( $p=0.007$ ). Myopia was the most common abnormality that was present in 5 patients (31.3%). The results showed that hyperopia was found in 2 patients (12.5%), squint in 2 patients (12.5%), retinitis pigmentosa 2 patients (12.5%), and blepharitis in 1 (6.3%) patient. Four other patients had multiple abnormalities: one with myopia and astigmatism, one with hyperopia and conjunctivitis, one with bilateral blepharitis, and allergic conjunctivitis, and the last one with myopia and blepharitis. Two children on examination of the eyes was diagnosed with Usher syndrome.

**Conclusion:** Ocular abnormalities are a common problem in children with congenital SNHL necessitating ophthalmological examination to detect any abnormal visual acuity, and aid in the diagnosis of congenital deafness syndromes.

*Saudi Med J 2009; Vol. 30 (9): 1197-1201*

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*Received 1st June 2009. Accepted 11th August 2009.*

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Hearing loss is the most common sensory deficit in humans.<sup>1</sup> Severe to profound bilateral deafness with early age of onset in children will greatly interfere with speech development, and also cognitive and psychological performance.<sup>2</sup> The incidence of severe hearing loss in newborn is 1/1000 live births, a rate that increases considerably in the case of children with risk factors for hearing loss.<sup>3</sup> At least 50% of cases of profound hearing loss can be attributed to genetic causes. Of these, 30% are associated with other disorders and thus, are referred to as cases of syndromic hearing loss. The remaining 70% are cases of non-syndromic hearing loss, up to 80% of which result from autosomal recessive inheritance, 10-20% from autosomal dominant inheritance, and 2-3% from x-linked inheritance.<sup>4,5</sup> A deaf person who is accustomed to heavy dependence on vision for information processing and communication can experience significant difficulties in communicating when a visual impairment occurs.<sup>6</sup> So in deaf children, early identification of visual deficits and prompt intervention are necessary for acquisition of information, cognition, social, and emotional development. As deaf children are mainly reliant on visual sensory input to overcome the loss of auditory input, it is vital to establish normal visual acuity.<sup>7</sup> Previous studies<sup>8,9</sup> reported that ocular abnormalities among deaf are higher than those children with normal hearing. There are 2 studies<sup>10,11</sup> investigating the ocular abnormalities among general pediatric population. The first study reported that 2.5% of American children had ocular deficits, while, the second study by the Baltimore Vision Screening Project noted that 14% of children had visual abnormalities. There is a known relationship between sensorineural hearing loss (SNHL) and vestibular hypofunction, which is another reason to have their vision tested (such as, if a child has SNHL, vestibular hypofunction and visual problems, then their balance and gaze stability will be greatly affected). The aim of the study is to evaluate the importance of ophthalmological examination in children with congenital SNHL.

**Methods.** This prospective study was conducted at the Ear, Nose, and Throat (ENT) Department in Al-Ramadi Teaching Hospital, Al-Ramadi city, Iraq, from 20 December 2007 to 30 October 2008. Fifty patients (18 years or younger) with unknown SNHL, recruited from the ENT Outpatient Department were included in the study. Twenty of them had established diagnosis of congenital SNHL, while the remaining 30 patients were examined for the first time. Diagnosis of congenital SNHL was based on a combination of: 1. any child presented with deafness, deaf-mutism, delayed speaking and/or delayed learning since birth, 2. any possible known causes of SNHL

were excluded by proper history taking from parents of the kids including: maternofetal infection such as rubella, perinatal complications, meningitis, mumps, prenatal or postnatal drug ototoxic effects, kernicterus, acoustic trauma, cerebral palsy, and any neurological disorders that might cause central visual problems, 3. if complete ENT and neurological examination revealed no possibility of conductive deafness 4. the diagnosis of SNHL is confirmed by the results of pure tone audiogram and auditory brainstem response test. The information collected for each patient included age at diagnosis, gender, family history of congenital SNHL, whether hearing loss was unilateral or bilateral (symmetrical or asymmetrical), and the severity of deafness. The patients were divided into 3 age groups: prelingual (<2 years), perilingual (2-4 years), and postlingual (>4 years). The severity of hearing loss suffered from by the patients was classified according to the recommendation of the British Society of Audiology.<sup>12</sup> The patients were screened ophthalmologically by slit lamp examination, to assess the anterior segment (eyelids, conjunctiva, cornea, anterior chamber, lens, and anterior part of the vitreous), and by using 1% cyclopentolate to achieve good dilatation for the assessment of the fundus by using the direct and indirect ophthalmoscope and +78 Volk lens (Volk Optical Inc., Mentor, Ohio, USA), which is used with a slit lamp. The cyclopentolate will provide cycloplegia to eliminate accommodation that may alter the real refractive error in the patient. Too young or uncooperative children were examined under general anesthesia, and their retina examined by direct and indirect ophthalmoscopy after cycloplegic refraction. Electroretinography was recorded for those children who were found to have pigmentary retinopathy during ophthalmological assessment. This study was approved by the Surgical Department, College of Medicine, Anbar University, Al-Ramadi, Iraq.

Chi square test was used for statistical analysis for comparing the results of the patients, with or without ocular abnormalities. The data was analyzed using SPSS version 12.0.

**Results.** A total of 50 patients, 18 years or younger with congenital SNHL was identified; 30 (60%) were male and 20 (40%) were female. The age range was 1-18 years, with a mean age of  $6.45 \pm 4.57$  years. The mean age for each group is: prelingual  $1.17 \pm 0.29$ , perilingual  $2.41 \pm 0.71$  and postlingual  $9.27 \pm 3.79$  years. The highest age group affected was postlingual age group, while the lowest was prelingual age group (Table 1). Demographic characteristics of patients with SNHL are shown in Table 2. The majority of ocular findings were found in the postlingual age group (13 [81.3%]) which is statistically significant. There was no statistically

**Table 1** - Age group and gender relationship.

Age group (years)	Male n (%)	Female n (%)	Total
Prelingual (0-<2)	1 (2)	2 (4)	3 (6)
Perilingual (2-4)	10 (20)	7 (14)	17 (34)
Postlingual (4>-18)	19 (38)	11 (22)	30 (60)
<b>Total</b>	<b>30 (60)</b>	<b>20 (40)</b>	<b>50 (100)</b>

**Table 2** - Demographic characteristics of 50 patients.

Characteristics	With ocular abnormalities* n (%)	Without ocular abnormalities† n (%)	P-value
<i>Age, years</i>			
Prelingual (0-<2)	1 (6.3)	2 (5.9)	1
Perilingual (2-4)	2 (12.5)	15 (44.1)	0.00004
Postlingual (4>-18)	13 (81.3)	17 (50)	0.007
<i>Gender</i>			
Male	11 (68.8)	19 (55.9)	0.245
Female	5 (31.3)	15 (44.1)	0.169
<i>Positive family history of congenital SNHL</i>	5 (31.3)	14 (41.2)	0.292
<i>Deafness</i>			
Unilateral	1 (6.3)	0	
Bilateral symmetrical	13 (81.3)	31 (91.2)	0.494
Bilateral asymmetrical	2 (12.5)	3 (8.8)	0.513
<i>Severity</i>			
Mild	0	0	
Moderate	2 (12.5)	2 (5.9)	0.108
Severe	3 (18.8)	8 (23.5)	0.445
Profound	11 (68.8)	24 (70.6)	0.866
*N=16, †N=34, SNHL - sensorineural hearing loss, p-values was calculated using the $\chi^2$ test			

**Table 3** - Ocular findings in children with sensorineural hearing loss in relation to age groups.

Ocular findings	Prelingual	Perilingual	Postlingual	Total
	n (%)			
Myopia	0	0	5 (31.3)	5 (31.3)
Hyperopia	0	1 (6.3)	1 (6.3)	2 (12.5)
Squint	0	1 (6.3)	1 (6.3)	2 (12.5)
Retinitis pigmentosa	1 (6.3)	0	1 (6.3)	2 (12.5)
Blepharitis	0	0	1 (6.3)	1 (6.3)
Myopia + astigmatism	0	0	1 (6.3)	1 (6.3)
Hyperopia + conjunctivitis	0	0	1 (6.3)	1 (6.3)
Bilateral blepharitis + allergic conjunctivitis	0	0	1 (6.3)	1 (6.3)
Myopia + blepharitis	0	0	1 (6.3)	1 (6.3)
<b>Total</b>	<b>1 (6.3)</b>	<b>2 (12.5)</b>	<b>13 (81.3)</b>	<b>16 (100)</b>

significant difference between patients with, or without ocular findings regarding the laterality, severity of deafness, and whether deafness is symmetrical or asymmetrical. The gender and positive family history of congenital SNHL of the patient showed no statistically significant value between the 2 groups. A total of 16 (32%) had ocular abnormalities. Myopia was the most common abnormality, which was present in 5 patients. Hyperopia in 2, squint was found in 2 patients, 2 patients with retinitis pigmentosa, and blepharitis in 1 patient. Four other patients had multiple abnormalities: one with myopia and astigmatism, one with hyperopia and conjunctivitis, one with bilateral blepharitis and allergic conjunctivitis, and the last one with myopia and blepharitis. Ophthalmological examination in 2 children with profound bilateral SNHL contributed to the diagnosis of hearing loss syndrome and retinitis pigmentosa, and was diagnosed as having Usher syndrome (Table 3).

**Discussion.** Dual hearing loss and visual loss is a dangerous combination. In children with SNHL, early ophthalmological examination is of vital role in the achievement of 2 purposes. The first purpose is to identify visual acuity, and to diagnose visual defects necessitating early intervention. Correction of visual acuity, as well as early intervention of other ocular abnormalities is of great value in maximizing visual sensory input, which is necessary for the development of these children who already have auditory deficits. The second purpose is to determine or confirm congenital deafness syndromes that are associated with eye abnormalities. The benefits from early diagnosis of syndromes are to give comfort to the kids and their families on the diagnosis, may be able to identify other syndromes related abnormalities, and to help them in genetic counseling.<sup>7</sup> Vestibular symptoms is not a typical feature of hereditary hearing impairment. However, if a patient reports dizziness or balance problems, functional testing of peripheral vestibular system in addition to ophthalmological examination should be performed. For instance, absent vestibular response can be seen in Usher syndrome Type 1, and in some forms of autosomal recessive non-syndromic hearing loss (such as, DFNB4, and others).<sup>1</sup>

Previous studies investigating ocular abnormalities in children with SNHL have reported prevalence ranging from 12-61%.<sup>7-9,13-19</sup> The variability in these studies may be attributed to differences in the age of the patient, patient population, place of the study (clinic versus institution), and the definition of what is abnormal. The prevalence of ocular findings noted in the present study is within the range reported in the literature. The association of the high frequency of ocular abnormalities in the deaf patients in our study and these studies is not

clearly understood. Anyhow, every child with SNHL should be sent for ophthalmological consultation to detect and treat ocular deficits in order to improve the quality of life of a deaf individual.

In the present study, the incidence of ocular findings was statistically not significant regarding laterality (unilateral versus bilateral) or severity (moderate, or severe, or profound) of SNHL, which is consistent with a previous study.<sup>13</sup> Furthermore, we found that ocular findings were not significantly affected by gender, positive family history of congenital SNHL, and symmetry of deafness (symmetrical versus asymmetrical). Although audiometric tests can be helpful in the evaluation of SNHL, they do not seem to be informative with respect to ophthalmological abnormalities. In the present study, ocular deficits were found to be higher in the postlingual age group than any other age groups, which is statistically significant ( $p=0.007$ ), this may be explained that the ocular abnormalities, in particular refractive errors, might be acquired during lifetime. This finding supports the idea that periodic ophthalmological examination is of utmost importance in the assessment and early treatment of deaf children.

Unfortunately in Iraq, there is no hearing screening program for newborns, even for those with risk factors, and because of this many deaf children during the study period were missed. Hearing screening program for all newborn allows detection of congenital and early onset hearing loss, for optimal acquisition of speech, learning, and psychological development.<sup>20</sup> It is our hope in the near future that our country adopts policies for mandatory newborn hearing screening program, thereby visual screening is required for deaf newborn to minimize their disabilities, and improvement in their quality of life. In this study, refractive errors were found in 10 (62.5%) patients which is consistent with other studies.<sup>7,8,14,18</sup> So, refractive error in our study is the most common ocular abnormality in children with SNHL.

Sharma et al's<sup>13</sup> study reported that non-refractive abnormalities were more than refractive conditions, which is inconsistent with our finding. The gap junction  $\beta$ -2 (GJB2) encodes a gap junction protein, connexin 26 (CX26), which is expressed in the inner ear, and is thought to be important in maintaining endocochlear potential.<sup>21</sup> Genetic testing for its mutations has the advantage for genetic diagnosis and counseling of autosomal recessive non-syndromic hearing loss. Recent studies<sup>22-25</sup> reported that the incidence of CX26 mutations among the deaf with autosomal recessive non-syndromic hearing loss was 26-50%. Sharma et al's<sup>13</sup> study reported 3.7% of the deaf children with ocular abnormalities had CX26 mutations.

As there is no hearing screening program for newborns for the time being in our country, the small sample size on the prelingual age group, has limited our study.

In conclusion, routine ophthalmological examination is of utmost importance in the evaluation and rehabilitation of deaf children. Future studies must include newborns with SNHL to be screened ophthalmologically after the establishment of the mandatory hearing screening program to help deaf children, as much as we can.

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#### Related topics

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