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Analysis of Newborn Hearing Screening Test Results of Children with Down Syndrome

Fatih Yüksel[1], Cüneyt Uğur[2]

[1]Department of Otorhinolaryngology, Konya City Hospital, Konya, Turkey
[2]Department of Pediatrics, University of Health Sciences Turkey, Konya Health Application and Research Center, Konya, Turkey

What is already known on this topic?

- *Individuals with Down syndrome (DS) are prone to ear, nose, and throat anomalies that cause hearing impairment. Thanks to neonatal hearing screening programs, it is possible to confirm the presence of congenital hearing loss in newborns with DS. Detecting hearing loss early and making the necessary interventions positively affects the child's language, social, and academic development.*

What this study adds on this topic?

- *It has been determined that infants with DS who do not have any risk factors for hearing loss have a significantly greater degree of hearing loss than healthy infants. From the results of the second screening test, no significant difference was found between the DS babies who failed and passed the test in terms of birth weight, gestational week, and maternal age.*

ABSTRACT

Aim: The aim of this study was to analyze newborn hearing screening test results of children with Down syndrome (DS).

Methods: The files of 84 children with DS and 84 healthy children (control group) admitted to the Pediatrics Polyclinics of the Konya Training and Research Hospital between January 2017 and June 2020 were retrospectively reviewed.

Results: Thirty-one of the 84 babies with DS were female (36.9%), and 53 were male (63.1%); 37 of the 84 babies in the control group were female (44%), and and 47 were male (56%) (P = .346). Fortty-eight (57.1%) of the 84 babies with DS and 17 (20.2%) of the 84 babies in the control group failed the first screening test (P < .001). It was determined that 24 (50%) of the 48 infants with DS who failed the first test also failed the second test and were referred, and all 17 infants in the control group who failed the first test passed the second test (P < .001). There was no significant difference in terms of birth weight, gestational week, and maternal age between infants with DS who failed and passed after the second screening test (P > .05 for all).

Conclusion: Our study shows that birth weight, gestational age, and maternal age do not pose an additional risk for hearing loss in DS babies who do not have known risk factors for hearing loss.

Keywords: Down syndrome, infant, hearing loss, newborn hearing screening

INTRODUCTION

DS is the most common anomaly among numerical chromosomal anomalies. It is caused by excess genetic material on chromosome 21 and its incidence is between 1/600 and 1/800.[1] DS is characterized by developmental delay, characteristic facial features, and hypotonia. It is associated with many systemic disorders such as mental retardation, dysmorphic findings, congenital heart diseases, hematological diseases, neurological anomalies, autoimmune diseases, gastrointestinal and endocrine system diseases, and vision and hearing problems. Symptoms appear at varying frequency and severity according to patients and populations.[2]

Individuals with DS are prone to ear, nose, and throat anomalies that cause hearing impairment. Inner ear hypoplasia and dysplasia of the cochlea, cochlear nerve canal, lateral semicircular canal, and cochleovestibular nerve are among the causes of sensorineural hearing loss in individuals with DS.[3] Early diagnosis of infants with hearing loss can help mitigate such negative consequences, as hearing loss in children will lead to lifelong deficits in speech and language acquisition, poor academic performance, personal-social disharmony, and emotional difficulties. In addition, speech and language development will be much more seriously affected in DS children with mental disability due to hearing loss. In studies on hearing

Corresponding author:
Fatih Yüksel
✉kbbfatih@yahoo.com
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screening test results performed in the neonatal period, it was reported that infants with DS failed the test at a rate of 25.7-36%.[4-6]

Through the National Neonatal Hearing Screening (NNHS) program launched in our country on December 3, 2004, all infants are followed closely and the necessary treatment methods are applied without delay. When we examined the literature, we did not come across any study related to newborn hearing screening results of children with DS in our country so far. In this study, we aimed to analyze the hearing screening test results in children with DS, using the data obtained within the scope of the NNHS.

METHODS

Study Population

Two hundred ten children who were admitted to the pediatric outpatient clinics of the Konya Training and Research Hospital between January 2017 and June 2020, and previously diagnosed with DS clinically and cytogenetically, were included in the study. The control group consisted of healthy gender-matched infants. Files of both groups within the scope of NNHS were examined retrospectively. The data of 84 cases in the DS group and 84 cases in the control group who met the study criteria were recorded.

Gender, age (age at the time of the hearing screening tests), delivery method (normal delivery/cesarean section), birth weight, gestational week, maternal age, and newborn hearing screening test results were recorded for both groups. The exclusion criteria for both groups were the toxoplasma, rubella, cytomegalovirus, and herpes (TORCH) group of infections, consanguineous marriage, familial history of hearing loss, premature birth (≤37 weeks), low birth weight (<2500 g), neonatal hyperbilirubinemia, craniofacial anomaly, history of hospitalization in the intensive care unit, ototoxic drug use, and any disease that could cause hearing loss.

Ethical Approval

This study complied with the ethical principles of the Declaration of Helsinki, and was approved by the Necmettin Erbakan University Faculty of Medicine Ethics Committee (Date: January 22, 2021, No: 2021/3055).

First Newborn Hearing Screening Test Method

The "Automatic Auditory Brainstem Response" (AABR) test (MADSEN, AccuScreen, Denmark) was administered to all patients as the first screening test after birth. Each patient underwent a routine otoscopic examination before each test. The infants' ears were cleaned with a cerumen in the external ear canal. All infants were evaluated bilaterally while sleeping without any sedation. The test result was reported as "passed" and "failed." Based on the results of the first test, those who passed through both ears were accepted as "passed." Those who failed in 1 or both ears were accepted as "failed."

Second Newborn Hearing Screening Test Method

Infants who failed the first test were recalled for the second test 2 weeks later and the AABR test was repeated. Tympanometric evaluation and acoustic reflex measurements were performed to provide information about the presence of fluid in the middle ears, the condition of the eardrum, middle ear pressure, and acoustic reflex of infants who failed the test. In the second AABR test, infants who received a "passed" response were deemed to have normal hearing levels. Those who failed the AABR test were referred to a secondary center for diagnostic testing.

Statistical Analysis

Data were analyzed using Statistical Package for the Social Sciences (SPSS) version 22.0 (IBM SPSS Corp.; Armonk, NY, USA). Descriptive statistical methods were used in the analysis of the data. Normality tests including the Kolmogorov–Smirnov and Shapiro–Wilk tests were performed to determine the distribution of data. Normally distributed data were expressed as mean ± standard deviation, and data not normally distributed were expressed as median (25th-75th percentile). Categorical variables were specified as number (*n*) and percentage (%). Comparison of numerical data between groups was made with the Student's *t*-test or the Mann–Whitney *U*-test. The chi-square test was used to compare categorical variables. A *P* value of less than .05 was considered statistically significant.

RESULTS

Clinical Features

All infants with DS were cytogenetically determined to have trisomy 21. The median age of the infants with DS at the time of the first screening test was 21 (15-27.8) days, and the median age of the infants in the control group was 21 (15-30) days. There was no significant difference between the ages of both groups ($P = .080$). While 31 of the 84 infants (36.9%) with DS were female, 53 were male (63.1%); 37 of the 84 infants (44%) in the control group were female and 47 (56%) were male ($P = .346$) (Table 1).

First Newborn Hearing Screening Test Results: It was determined that infants with DS had statistically significant lower birth weight (2890 (2682.5-3122.5) g vs. 3192.5 (2950-3537.5) g, respectively, $P < .001$), lower gestational week (38 (38-39) weeks vs. 39.5 (38-40) weeks, $P < .001$), and higher maternal age (35 (29-40) years vs. 28 (24-31) years, respectively, $P < .001$) compared to the control group. There was no significant difference between the 2 groups in terms of delivery method ($P = .089$) (Table 1). In the first screening test, 48 (57.1%) of the 84 infants with DS and 17 (20.2%) of the 84 infants in the control group failed the test. The rate of failing from the first test was significantly higher in infants with DS compared to the control group ($P < .001$) (Table 1).

Second Newborn Hearing Screening Test Results: At the time of the second screening test, the median age of 48 infants with DS was 36 (36-42) days, and the median age of 17 infants in the control group was 42 (36-51) days. There was no significant difference between the 2 groups in terms of age ($P = .104$) (Table 2). It was observed that male infants with DS remained significantly higher in the second test ($P = .029$). Infants with DS, who had a second screening test, were found to have significantly lower birth weight (2865 (2690-3085) g vs. 3200 (3100-3420) g, respectively, $P = .001$), lower gestational week (38.5 (38-39) weeks vs. 40 (38.5-40) weeks, respectively, $P = .015$) and higher maternal age (34.3 ± 6.8 years vs. 26.8 ± 3.7 years, respectively, $P < .001$) than the control group

Table 1. Distribution of the Demographic, Pregnancy, and First Hearing Screening Test Data of Both Groups

Parameters	Down Syndrome ($n = 84$)	Control ($n = 84$)	*P*
Age (at the time of the first screening test) (days)*	21 (15-27.8)	21 (15-30)	.080[α]
Gender#			
Female	31 (36.9)	37 (44)	.346[β]
Male	53 (63.1)	47 (56)	
Birth weight (g)*	2890 (2682.5-3122.5)	3192.5 (2950-3537.5)	<.001[α]
Gestational age (weeks)*	38 (38-39)	39.5 (38-40)	<.001[α]
Delivery method#			
Vaginal	39 (46.4)	50 (59.5)	.089[β]
Cesarean	45 (53.6)	34 (40.5)	
Mother's age (years)*	35 (29 - 40)	28 (24-31)	<.001[α]
First screening test#			
Passed	36 (42.9)	67 (79.8)	<.001[β]
Failed	48 (57.1)	17 (20.2)	

*Values were expressed as median (25th-75th percentile); [α]*P* values for Mann–Whitney *U*-test; #Values were expressed as *n* (%); [β]*P* values for chi-square test.

infants. There was no significant difference between the groups in terms of delivery method ($P = .722$). It was determined that 24 (50%) of 48 infants with DS who failed the first test failed the second test and were referred, and all 17 infants in the control group who remained from the first test passed the second test. The rate of failing the second test was found to be significantly higher in babies with DS compared to the control group ($P < .001$) (Table 2).

Analysis of the Data of Babies with DS who Failed and Passed the Second Newborn Hearing Screening Test: There was no significant difference between the DS babies who passed the second test and those who failed, in terms of age, gender, and delivery method (*P* values; .103, 1.000, and .773, respectively). While it was observed that the infants with DS who failed the second screening test had lower birth weight, lower gestational age, and higher maternal age compared to those who passed, it was observed that there was no significant difference between both groups in terms of these 3 parameters ($P > .05$ for all) (Table 3).

DISCUSSION

Most of the children with DS frequently apply to an otolaryngologist for medical and surgical treatment due to recurrent ear infections and possible hearing loss problems from the first years of their life.[7] It has been reported that the rate of hearing loss ranges from 38% to 78% in all DS patients.[4,5] Hearing loss studies in individuals with DS are studies that generally involve children and adults and report hearing loss rates calculated using different hearing test methods. It has also been shown that these individuals have early-onset presbycusis, and the frequency of sensorineural hearing loss increases with age.[4,8] For these reasons, it is also important to determine the existing hearing loss at birth by neonatal testing in these individuals, who are already predisposed to problems with hearing at advanced ages.

Basonbul et al.[6] reported that 36% of infants with DS failed in newborn hearing screening and only 9% of these patients had normal hearing in subsequent tests. Park et al.[5] reported

Table 2. Distribution of the Demographic, Pregnancy, and Screening Test Data of Infants Who Underwent Second Hearing Screening Test in Both Groups

Parameters	Down Syndrome ($n = 48$)	Control ($n = 17$)	*P*
Age (at the time of the second screening test) (days)*	36 (36-42)	42 (36-51)	.104[α]
Gender#			
Female	14 (29.2)	10 (58.8)	.029[β]
Male	34 (70.8)	7 (41.2)	
Birth weight (g)*	2865 (2690-3085)	3200 (3100-3420)	.001[α]
Gestational age (weeks)*	38.5 (38-39)	40 (38.5-40)	.015[α]
Delivery method#			
Vaginal	23 (47.9)	9 (52.9)	.722[β]
Cesarean	25 (52.1)	8 (47.1)	
Mother's age (years)[Ω]	34.3 ± 6.8	26.8 ± 3.7	<.001[δ]
Second screening test#			
Passed	24 (50)	17 (100)	<.001[β]
Failed	24 (50)	0 (0)	

*Values were expressed as median (25th-75th percentile); [α]*P* values for Mann–Whitney *U*-test; [Ω]Value was expressed as mean ± standard deviation; [β]*P* values for chi-square test; #Values were expressed as *n* (%); [δ]*P* value for Student's *t*-test.

Table 3. Distribution of the Demographic and Pregnancy Data of DS Group Who Passed and Failed from Second Hearing Screening Test

Parameters	Passed ($n = 24$)	Failed ($n = 24$)	*P*
Age (at the time of the second screening test) (days)*	36 (31.5-40.5)	36 (36-49.5)	.103[α]
Gender#			
Female	7 (29.2)	7 (29.2)	1.000[β]
Male	17 (70.8)	17 (70.8)	
Birth weight (grams)*	2900 (2692.5-3073.8)	2825 (2622.5-3178.8)	.695[α]
Gestational age (weeks)*	39 (38-39)	38 (38-39)	.290[α]
Delivery method#			
Vaginal	12 (50)	11 (45.8)	.773[β]
Cesarean	12 (50)	13 (54.2)	
Mother's age (years)[Ω]	34.1 ± 6.8	34.4 ± 6.9	.883[δ]

*Values were expressed as median (25th-75th percentile); [α]*P* values for Mann–Whitney *U*-test; #Values were expressed as *n* (%); [β]*P* values for chi-square test; [Ω]Value was expressed as mean ± standard deviation; [δ]*P* value for Student's *t*-test.

that 87 (26.2%) of 332 patients with DS that they included in the study could not pass newborn hearing screening. They suggested that more than 43% of children who passed neonatal hearing screening developed conductive hearing loss requiring insertion of ventilation tubes and that DS children should be seen for follow-up every 3-6 months. Tedeshi et al.[4] in their study on newborns with DS, reported that 28 (25.7%) of 109 patients with hearing screening data failed in the neonatal hearing screening test, and after the subsequent tests, the prevalence of congenital hearing loss in newborns was 15%. Considering these studies, the rate of congenital hearing loss in children with DS varies between 15% and 36%. In accordance with the NNHS program implemented in our country, all newborns are given postnatal hearing screening tests. In our study, the rate of failing the first test in infants with DS was 57.1%, and the rate of failing the second test in babies who failed the first test was 50%. Altogether 24 (28.6%) of 84 DS infants included in our study were referred to the advanced center after failing the second test. However, the final diagnosis of the referred infants could not be reached due to the distribution of the infants in different centers and because they were not included in the registries of the NNHS.

The results of neonatal screening programs, in a study by Bolat and Genç[9] in Turkey, showed that 5485 babies were screened, 11 babies (0.2%) were diagnosed with bilateral severe hearing loss. Again, according to NNHS results in Turkey, Çelik et al.[10] reported the rate of failing the first test as 18.77%, the rate of failing the second test as 1.69%, and ultimately the rate of hearing loss as 0.27%. In our study, although 17 (20.2%) of 84 healthy infants failed the first test, all of the remaining babies passed the second test.

A family history of hearing loss in infants, consanguineous marriage, TORCH group infections, preterm birth (≤37 weeks), low birth weight (<2500 g), neonatal hyperbilirubinemia, craniofacial anomaly, history of hospitalization in intensive care for more than 5 days, and use of ototoxic drugs for hearing loss are reported as risk factors.[11,12] Connolly et al.[13] found the rate of hearing loss to be 0.01% in infants with no risk factors, and 0.1% in infants with risk factors. Sarbay et al.[14] reported from their study that while 17% of the babies in the group with risk factors failed the first test, 29.4% of the remaining babies also failed the second test. As the number of risk factors increases, the rate of failing from the ABR test increases significantly.[14] In our study, those with risk factors that may cause hearing loss, both healthy infants and those with DS, were excluded from the study.

According to the Joint Committee on Infant Hearing and the Healthcare Supervision for Children with DS Guidelines, follow-up audiologic assessments should be completed by the age of 3 months.[15,16] In our study, the median age of the infants with DS at the time of the first screening test was 21 (15-27.8) days, and the median age of the infants in the control group was 21 (15-30) days. At the time of the second screening test, the median age of infants with DS was 36 (36-42) days, and the median age of infants in the control group was 42 (36-51) days. In our study, the first and second tests in both groups were done before the infants were 3 months old.

In our study, while it was observed that the infants with DS who failed the second screening test had lower birth weight, lower gestational age, and higher maternal age compared to those who passed, there was no significant difference between both groups in terms of these 3 parameters. There was no significant difference between the DS babies who passed the second test and those who failed in terms of age, gender, and delivery method. As a result, it was seen in our study that birth weight, gestational age, and maternal age do not pose an additional risk for hearing loss in DS babies who do not have known risk factors for hearing loss.

Study Limitations

Our study was retrospective, and performed at a single center with a relatively small patient and control population, and these factors were considered the limitations of our study. In addition, the AABR test performed in our institution in line with the NNHS program is a method that can miss detection of hearing loss at a threshold of 35 dB and below. Multicenter prospective studies with a larger sample size and longer follow-up can help determine the frequency of hearing loss in children with DS and risk factors affecting hearing, as well as the type, timing and frequency of the hearing test.

CONCLUSION

In our study, it was found that babies with DS who did not have any risk factors for hearing loss had significantly more hearing loss than healthy babies. Therefore, since DS by itself is a risk factor for hearing loss, DS babies should be closely monitored and examined in detail in terms of hearing assessment, starting from the neonatal period. In addition, we think that multicenter studies with a large number of patients will be useful to determine the differences between DS babies with and without hearing loss.

Ethical Committee Approval: Ethics committee approval was received for this study from the ethics committee of Necmettin Erbakan University Faculty of Medicine (Date: January 22, 2021, No: 2021/3055).

Informed Consent: Informed consent was not obtained due to the retrospective design of this study.

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