





30th Congress of Union of the European Phoniatricians

April 27 - 30, 2023
Xanadu Convention Center
Antalya - TÜRKİYE

ABSTRACT BOOK

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ORGANIZING SECRETARIAT
Topkon Congress & Event Management
Phone: +90 216 330 90 20
E-mail : uep2023@topkon.com

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ORAL PRESENTATIONS

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151-MOLECULAR GENETIC ASPECTS OF SENSORINEURAL HEARING IMPAIRMENT

Gavkhar KHAYDAROVA[1], Khalida Shaykhova[1], Aziza Madrimova[2]
[1]Tashkent Medical Academy
[2]Urgench branch of Tashkent Medical Academy

Objectives: The study of clinical and molecular genetic aspects of the development of hearing loss in newborns. **Methods:** The study included 454 children born from 2008 to 2012 with bilateral sensorineural hearing loss diagnosed in the first year of life based on the results of a detailed audiological examination. The examined group included children aged 3 weeks to 11 months. The average age at the time of the survey was 6 months, the sex ratio was 1:1 (233 boys and 221 girls). When examined by a geneticist, the presence of clinical data on the syndromic nature of the disease was excluded. All children underwent a study of the GJB2 gene. The dynamic observation group included 2 13 children who underwent repeated audiological examinations with an interval of 3-4 months, which made it possible to assess the nature of changes in hearing thresholds over time. **Results:** As a result of a genetic examination of 454 infants with bilateral sensorineural hearing loss, the genotype with pathological mutations was detected in 226 children (49.7 %), and the genotype with two mutations was found in 201 (42.2 %) cases, with one mutation (heterozygous genotype) - in 19 (4, 18 %) cases. The genotype without mutations was found in 81 (18.0 %) children **Conclusions**: The identification of two mutations in the patient's genotype indicates a hereditary cause of hearing loss. In the remaining 8 1 children with an unchanged genotype, one could assume some cause of the deafness. In genotype 19 (4.18 %) children, only one mutation was found, which corresponds to the genotype of a healthy carrier, and in 6 cases it was the c.35delG mutation. The study demonstrates a significant contribution of pathogenic mutations of the GJB2 gene to the etiological structure of bilateral sensorineural hearing loss (49.7%).

Keywords: genetic, hearing loss, molecular

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