



GENETIC ASPECTS OF SENSORINEURAL HEARING IMPAIRMENT IN CHILDREN

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Annotation

Auditory perception is the result of the joint activity of the auditory perceiving and brain analyzing and integrating systems. As the final link in the line of acoustic communication, hearing, as well as the specific behavior caused by it, is controlled and coordinated by many complex brain mechanisms. The study of clinical and molecular genetic aspects of the development of hearing loss in newborns.

Material and research methods . The study included 454 children born from 2018 to 2023 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 213 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. Conclusion. The identification of two mutations in the patient's genotype indicates a hereditary cause of hearing loss. In the remaining 81 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%).

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