ETIOLOGICAL PROFILE OF HEARING LOSS IN PEDIATRIC COCHLEAR IMPLANT USERS

Jekaterina Byčkova1,2, Gabriele Černytė3, Margarita Gromova3, Silvija Kiverytė4,5, Violeta Mikščienė4, Eugenijus Lesinskas1

1. Clinic of Ear, Nose, Throat, and Eye Diseases, Institute of Clinical Medicine, Faculty of Medicine, Vilnius University
2. Children’s Otolaryngology and Ophthalmology Department, Children’s Hospital, Affiliate of Vilnius University Hospital Santaros Klinikos
3. Faculty of Medicine, Vilnius University
4. Department of physiology, biochemistry, microbiology and laboratory medicine, Institute of Biomedical science, Faculty of Medicine, Vilnius university
5. Centre of Laboratory Medicine, Vilnius University Santaros Klinikos
6. Department of Human and Medical Genetics, Institute of Biomedical Sciences, Faculty of Medicine, Vilnius University

Introduction
Congenital sensorineural hearing loss (HL) is a heterogeneous disorder, its etiologic profile varies between populations. Pathogenic variants of GJB2 gene are the major causes of non-syndromic HL. Congenital cytomegalovirus infection (cCMV) is the most important prenatal etiologic factor causing HL and other lesions. Perinatal events, syndromes, infections or traumas are less common. The causes of the remaining one third of HL cases are unknown.

Aim of the study:
To determine the etiologic profile of HL in deaf children cochlear implant users.

Materials and methods:
The data of 123 children with cochlear implants were analysed. Sequencing of the genes associated with HL was performed to 81 children. CMV DNA was extracted from DBS on Guthrie cards and detected using RT-PCR to 109 children. Retrospective cCMV analysis was based on DBS, initially obtained during the first 3-5 days of neonate life for the Universal screening of the inherited metabolic diseases. In order to enhance sensitivity, DNA was extracted in triplicates. A DBS 5 mm in diameter were used for a single DNA extraction. DBS samples were considered positive when two or more of the triplicate PCR reactions tested CMV positive. Patients’ medical records were analysed for identification of other etiologic factors.

Results
GJB2 gene pathogenic variants were determined to 45 individuals (37 % of the study group), additionally alterations in other genes causing non-syndromic HL were identified to 5 (4 %) participants. 8 children (7 %) were diagnosed with syndromic HL (3 cases of Pendred syndrome, 2-Usher, 1-Rogers, 1 Jacobsen, 1-CHARGE). CMV DNA was revealed in 12 (10 %) samples. Perinatal and postnatal (e.g. meningitis and trauma) events caused HL to 17 (14 %) and 4 (3 %) children respectively. The cause of hearing loss remains unknown for 31 (25 %) children.

Conclusions
The major causes of HL in our study were GJB2 gene alterations followed by cCMV infection. The other factors were infrequent. HL of idiopathic origin remains to the quarter of all our study participants.

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Contact e-mail: jekaterina.byckova@gmail.com