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Phenotype and genotype characterisation of Usher syndrome in Russian cohort

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Background: Usher syndrome (USH) clinically shows retinitis pigmentosa and deafness. USH prevalence is 3,2-6,2/100,000. Four clinical sub-types are identified. Due to heterogeneous clinical picture, genetic test is recommended for precise diagnosis and effective treatment. Although the mutation spectrums of USH associated genes are reported in many populations, this is the first comprehensive study for Russian population.

Methods: 28 subjects with USH were selected from 3200 patients from Deaf-Blind Support Foundation "Con-nection" following observational study protocol NCT03319524. Examinations included visometry, perimetry, OCT, ophthalmoscopy, electroretinography, recording of visual-evoked potentials, tonal and electronic audiometry, acoustic impedance measurement, vestibulometry, electronystagmography, and posturometry. NGS, MLPA and Sanger sequencing were considered for genetic analysis.

Findings: 53.57% and 39.28% patients had USH1 and USH2, respectively. 73.33% subjects showed variations in USH1 associated genes MYO7A (77%), CDH23 (5,5%), PCDH15 (12%), and USH1C (5,5%). 11 mutations were detected in MYO7A where 54,54% are novel. MYO7A: p.Q18* was the most frequent (27,27%) mutation and found to be associated with early manifestation and most severe clinical picture. Two novel mutations (p.E1301* and c.158- ?_318+?del) were detected in PCDH15 gene. 90,90% patients suspected to be USH2 confirmed by genetic testing have shown 11 mutations in the USH2A gene, where 27,27% were novel. In 50% cases, we observed USH2A: p.W3955* with p.E767fs, p.R1653* and c.8682-9A>G (16,7% each).

Interpretation: The Russian USH cohort shows both novel and known USH mutations. Clinically the prevalence of USH2 is low (39.28%) and the frequency of MYO7A mutations responsible for USH1B is very high (81,81%) compared to other cohorts indicating that these nine USH1 patients might be eligible for UshStat® clinical trial. The diagnoses of USH1 and USH2 were genetically confirmed in 73.33% and 90.90% cases respectively, indicating the efficacy of our clinical protocol.

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Biography

Marianna Ivanova graduated from Moscow State University, got MD in ophthalmology, PhD in visual research. Chief of Oftalmic CRO and genetic testing company in ophthalmology, established in 2008, headquarter in Moscow. Main focus of the company – providing best quality clinical research and services for patients with inherited forms of ocular pathology..

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