Секция «Экспериментальные исследования»

Study of molecular mechanisms of FMF using sequencing and genetic engineering approaches.

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Familial Mediterranean fever (FMF) is an autoinflammatory disease that is prevalent in Mediterranean countries, including Armenia [1]. The disease is caused by mutations in the *MEFV* gene, which is responsible for encoding the pyrin protein and is located on the short arm of the 16th chromosome. This study aimed to perform whole-exome sequencing (WES) analysis for FMF patients in order to obtain comprehensive information about their genetic variability in coding regions of the *MEFV* gene. For this purpose, 4 patients diagnosed with FMF have been enrolled. Genomic DNA was extracted using a modified salting out approach. WES has been performed using commercially available services. For bioinformatic analysis of the data GATK tools were applied. Among other functional genetic variants, confirmed by the independent PCR-based molecular genetic testing approach, WES allowed identification of R202H functional genetic variant in one of the studied patients. Further analysis with the enrollment of additional samples is needed to clarify the role of other functional variants in disease pathogenesis. To assess the functional role of each genetic variant with unknown clinical significance we have developed pluripotent cell lines of healthy subjects and plan to further use genetic engineering approaches [2].

Источники и литература

1) 1. Omer F Beşer, Ozgür Kasapçopur, Fügen Cullu Cokuğraş, Tufan Kutlu, Nil Arsoy, Tülay Erkan. Association of inflammatory bowel disease with familial Mediterranean fever in Turkish children. DOI: 10.1097/MPG.0b013e31827dd763 2. Elena V. Grigor'eva, Anastasia A. Malakhova, Lilit Ghukasyan, Varduhi Hayrapetyan, Sofi Atshemyan, Valentina Vardanyan, Suren M. Zakian, Roksana Zakharyan, Arsen Arakelyan. Generation of three induced pluripotent stem cell lines (RAUi001-A, RAUi001-B and RAUi-001-C) from peripheral blood mononuclear cells of healthy Armenian individual. DOI: 10.1016/j.scr.2023.103147