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Personalized assessment of mitochondrial DNA (mitDNA) specific features

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Introduction: Collective term 'mitochondrial disease' means heterogeneous group of disorders caused by mutations in mitochondrial genes. There are several classical syndromes, such as MELAS, MERRF, Leigh etc. However, the majority of mitochondrial diseases does not have certain genotype-phenotype correlation. There are two major groups of mitochondrial variants. First includes all types of haplogroup polymorphisms (usually, 30-100 per person). Second consists of rare or novel variants with undefined significance (2-5 per person in average). Every variant from the second group could be important for disease development. Therefore, accurate assessment of mtDNA rare or novel variants is very important.

Methods: We researched group of 50 children with diagnosed non-syndromic mitochondrial encephalopathy. Samples of peripheral venous blood were used. Whole mitochondrial sequence was made with Sanger method. Obtained sequences were compared with rCRS, mitochondrial haplogroup was determined according to *PhyloTree*. All novel or rare variants were assessed *in silico*.

Results and Discussion: We evaluated none of confirmed mutations, associated previously with mitochondrial disease. However, we found 27 novel or rare (frequency <0,2%, hmtDB). 16 of them (59%) have high pathogenic potential. Special attention must be paid to variant A10732T, which was found in 10 not related patients. On the ground of haplogroup analysis – all 10 considered patients belonged to completely different subclades - we conclude that this variant has to play important role in development of mitochondrial encephalopathy.

Biography

Sergey Suchkov, MD, PhD was born in 11.01.1957, a researcher-immunologist, a clinician, graduated from Astrakhan State Medical University, Russia, in 1980. Suchkov has been trained at the Institute for Medical Enzymology, The USSR Academy of Medical Sciences, National Center for Immunology (Russia), NIH, Bethesda, USA) and British Society for Immunology to cover 4 British university facilities. Since 2005, Dr Suchkov has been working as Faculty Professor of I.M. Sechenov First Moscow State Medical University and Of A.I.Evdokimov Moscow State Medical & Dental University. From 2007, Suchkov is the First Vice-President and Dean of the School of PPPM Politics and Management of the University of World Politics and Law. In 1991-1995, Dr Suchkov was a Scientific Secretary-in-Chief of the Editorial Board of the International Journal "Biomedical Science" (Russian Academy of Sciences and Royal Society of Chemistry, UK) and The International Publishing Bureau at the Presidium of the Russian Academy of Sciences. In 1995-2005, Suchkov was a Director of the Russian-American Program in Immunology of the Eye Diseases. Dr Suchkov is a member of EPMA (European Association of Predictive, Preventive and Personalized Medicine, Brussels-Bonn), a member of the NY Academy of Sciences, a member of the Editorial Boards for Open Journal of Immunology and others. Dr Suchkov is known as an author of the Concept of post-infectious clinical and immunological syndrome, co-author of a concept of abzymes and their impact into the pathogenesis of autoimmunity conditions, and as one of the pioneers in promoting the Concept of PPPM into a practical branch of health services

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