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Mitochondrial DNA variation in atherosclerosis

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In human pathology, several diseases are associated with mutations in the mitochondrial genome (mtDNA). Genetic predisposition plays an important role amidst the other risk factors in the development of atherosclerosis, a socially significant multifactorial disease. Even though mitochondrial dysfunction leads to increased oxidative stress, the role of mitochondrial mutations in atherosclerosis has not received much attention so far.

Conclusions: The focality and mosaic character of atherosclerotic lesions in human aortic intima may be due to the differences in the heteroplasmy level of mtDNA mutations to a great extent, and a certain profile of pro and anti-atherosclerotic mutations of mitochondrial genome is characteristic for different types of atherosclerotic lesions. The presence of heteroplasmic non-synonymous mtDNA mutations may lead to mitochondrial dysfunction in specific sites of intimal tissue. The data obtained in clinical study can be used to assess individual risk of atherosclerosis, as well as for further studies on the role of mitochondrial genome mutations in the development of atherosclerosis and its clinical manifestations. The individual profile of certain mtDNA variants may partially explain atherosclerosis variability and genetic predisposition to atherosclerosis in population, which could be inherited by maternal line.

Biography

Igor A Sobenin has got his MD in 1988 (Chelyabinsk State Medical Institute, Russia), PhD in 1991 (Russian Endocrinology Research Center), and DSc in 2006 (Institute of General Pathology and Pathophysiology, Russia). Now, he is a Leading Researcher at Russian Cardiology Research and Production Complex (Moscow, Russia). His research activity is in a field of molecular and cellular mechanisms of atherosclerosis, genetic and phenotypic markers of susceptibility, clinical, epidemiological and population studies in the field of atherosclerosis. He has over 200 published papers, among them over 120 are in international peer-reviewed journals.

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