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Mithocondrial function impairments determined by mtDNA mutations

Dr. Luigi Donato

University Of Messina,Italy

Abstract

The aim of the study is to Mithocondrial function impairments determined by mtDNA mutations could unveil retinal degeneration unknown etiopathogenesis mechanisms.
Mitochondria are considered one of the most essential organelles of eukaryotic cells. The most intriguing aspect of mitochondria lies in the uniqueness of its own genome, the mtDNA. It is already known that a lengthy accumulation of lower levels of mtDNA damage and mtDNA copy reduction could be linked to etiopathogenesis of neurodegenerative and age-related diseases. Despite an actual number of about 15,000 reported variants, only a few hundred are confirmed as disease causing. Today, the development of next generation sequencing (NGS) techniques permits efficient analyses of mtDNA, improving sample output and sensitivity of variant detection. Main issues of mtDNA high-throughput sequencing deal with detection and interpretation of low heteroplasmy and homoplasmy levels, variants unrelated to exhibited phenotype, and identification of variants of unknown significance. We analyzed the mtDNAs raw data extracted from exomes (WES) belonging to patients affected by retinal degeneration orphan forms, proposing an integrated approach based on the complementary use of the most recent algorithms applied to mtDNA data. We found variants carried by genes involved in fundamental cellular activities, such as induction of apoptosis by release of cytochrome C and following caspase activation, storage of calcium ions, heat production by non-shivering thermogenesis, production of cellular ATP and establishment of membrane potential by oxidative phosphorylation. In this way, a higher quality output can be obtained, leading to improved genetic counseling for people affected by primary mitochondrial retinal diseases.

Biography:

Luigi Donato, PhD in "Applied Biology and Experimental Medicine", frequents the Labs of Molecular Genetics of University of Messina, Italy. He is a researcher of the IEMEST institute in Palermo, Italy, too. He published more than 40 papers in reputed journals and participated in more than 25 national and international congresses, also being in the Organizing Committee in three of them. He was a member of ARVO and he is a member of AIBG. He joined the Editorial Board of several journals, also acting as Guest Editor for "Antioxidants". His main research fields are retinal dystrophies and omics approaches.

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