

Novel mutations in ND3 and Cyt b genes associated with coronary artery disease

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Introduction: Mitochondrial disorders are a group of clinically heterogeneous diseases which result due to oxidative phosphorylation (OXPHOS) defects. Several studies have implicated the role of mitochondria in the development of type II diabetes by providing the evidence that mitochondrial function is crucial for the glucose-stimulated secretion of insulin by pancreatic beta cells. Till now, a number of mtDNA defects have been implicated in the development of diabetes in various population. Mitochondrial diabetes was reported to account for about 10 % of the T2D population.

Objective: To study the mitochondrial DNA (mtDNA) variations in a patient with triple vessel coronary artery disease.

Research Design and Methods: A 75 year old man with a history of 30 years of type 2 diabetes (T2D) presented with uncontrolled blood sugar, elevated level of CK and CK-MB, unstable blood pressure. He was diagnosed with triple vessel coronary artery disease. In addition to this, the patient also had symptoms of neuropathy and cataract.. The patient's family history revealed maternal inheritance of type II diabetes. The DNA isolated from the heart muscle tissue was subjected to complete mitochondrial genome analysis and the functional implications of the novel mutations were analysed using bio-informatic tools.

Results and Discussion: Analysis of the complete mitochondrial genome revealed 2 novel mutations viz A10188G and A14782C in the regions of ND3 and Cyt b respectively. Further in silico analysis of these missense mutations using Polyphen, SIFT and PMut predicted altered pattern of secondary structure and pathological function.

Biography

V Vijaya Padma received her Ph.D. from University of Madras, India for the work on "Biochemical studies on the effect of fish oil in isoproterenol induced myocardial infarction". She worked as a Lecturer in Kongu Nadu Arts and Science College, Coimbatore for three years. She joined the Department of Biotechnology, School of Biotechnology and Genetic Engineering, Bharathiar University, as a Lecturer in April 2005. She established Animal Tissue Culture and Molecular Genetics Laboratory apart from teaching the Post Graduate students. Now her research group is involved in testing various phytochemicals as cytoprotective agents against toxins-mycotoxins, pesticides, dioxin and also to evaluate their anticarcinogenic, antimutagenic and anti-inflammatory properties. They are also involved in understanding the role of mitochondrial mutations in various disease conditions. She has published several research articles in both national and international peer reviewed journals.

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