5th International Conference on
HUMAN GENETICS AND GENETIC DISEASES
&
11th International Conference on
GENOMICS AND PHARMACOGENOMICS
September 21-22, 2018 | Philadelphia, USA

Analysis of mitochondrial DNA D-loop region mutations in Iranian patients with non-alcoholic fatty liver disease

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Introduction: Non-alcoholic fatty liver disease (NAFLD), is the term for a range of conditions caused by excessive lipid accumulation in hepatocytes in the absence of excess alcohol intake or other pathological causes. NAFLD is now the most common liver disease in the world. Mitochondria are the main source of cellular reactive oxygen species (ROS) in cells and also play a key role in fatty acid ß-oxidation and oxidative phosphorylation and therefore mitochondrial dysfunction will lead to accumulation of free fatty acids. Generated mutations by ROS in the mitochondrial DNA D-loop region could disturb mitochondrial function, oxidative phosphorylation & ATP production. Therefore these data strongly suggest that NAFLD might be a mitochondrial disease. The aim of this study was to the analysis of mutations in the mitochondrial DNA D-loop region in NAFLD patients.

Methodology & Theoretical Orientation: Genomic DNA was extracted from fresh liver tissue samples of NAFLD patients and control subjects by using a DNA isolation kit. Two pairs of primers designed for PCR amplification to amplify the mtDNA D-loop region and subsequently were sequenced using a sequence analyzer. The results of the sequencing were confirmed with a human mitochondrial database which mtDNA mutations have been reported in a spectrum of clinical disorders.

Findings: After D-loop sequencing, 85 different variations including 3 deletions, 6 insertions and 76 single nucleotide polymorphisms (SNPs) were detected. A significant difference was seen between two groups in 6 variations ($P<0.05$; T334C, C16111T, A16220C, C16266T, c16221ins, A248del). A novel insertion (16221 ins C) was observed in patients.

Conclusion & Significance: we think that the disease has damaged the mitochondrial DNA and these mutations have been created. Our findings indicate that D-loop alterations are frequent in NAFLD patients and may play a significant role in the progression of NAFLD.

Biography

Kambiz Hasrak has graduated from Hamadan University of Medical Sciences, Iran in the field of molecular medicine (PhD). His thesis was about Investigation of primary or secondary origin of mitochondrial DNA variations in Colon cancer patients. Now, he is working on cancers and genetic diseases and also Thiamine-responsive megaloblastic anemia syndrome in a Molecular Genetics Laboratory in Baqiyatallah El-Azam Hospital in Tehran.

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