

هفدهمین کنفرانس سراسری و پنجمین کنفرانس بین المللی زیست شناسی ایران



The Novel Mitochondrial Heteroplasmic Mutation (m.9140 C>G) in an Iranian Family with Long QTS (LQT3)

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Abstract

Mitochondrial DNA (mtDNA) could be considered a candidate modifier factor for Syndrome of Long QT (LQTS), since mitochondrial oxidative stress is thought to be involved in ATP production. It has been reported that the activity of ion channels in cardiomyocytes is sensitive to ATP level. We searched 40% of the entire mitochondrial genome in an Iranian family with LQT3 for mutation by PCR-SSCP and DNA sequencing. We report four new mutations and one reported mutation, leading to an amino acid substitution and two mutations in mitochondrial tRNA. We found statistically significant correlation ($r = 0.737$) between QTc (ms) and age of LQTS patients. Our data suggest that these mitochondrial mutations in a family with LQTS might be responsible mitochondrial defects and increase the gravity of Syndrome of Long QT (LQTS).

Keywords: Arrhythmia, Long QT syndrome, Mitochondrial DNA, Mutation, SSCP.

Introduction

Inherited arrhythmogenic disorders, especially long QT (LQT) syndromes have been suggested as major causes of Sudden Cardiac Death (SCD) in young individuals and affect patients with anatomically normal hearts (1, 2). Long QT syndrome (LQTS) is caused by mutations in genes encoding sodium or potassium channels (4-7). Exercise, emotion, anger, startle and fright are the most common triggers for symptoms among LQT patients (1). As yet, most investigations have focused on abnormalities of nuclear genes in arrhythmia (genes of encoded subunits of ion channels), but they

explain 50% to 60% of clinically diagnosed cases (6).

MtDNA mutations have been known to be involved in a wide range of diseases and syndromes. Although the precise mechanism whereby mutations of mitochondrial DNA influence these diseases is unclear, they may play an important etiological role. Since heart is highly dependent on oxidative energy generated in mitochondria and