# *In silico* analysis of mitochondrial tRNA$^{lue(CUN)}$ structure in the presence of A12308G polymorphism in patients with non-dystrophic myotonia

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**Abstract:** Non-Dystrophic Myotonia is a rare disease with a prevalence of 1 per 100,000 people. This illness is an important group of skeletal muscle channel disorders that diagnosed by a change in the irritability of the membrane in terms of physiologically. The clinical symptoms of this condition include muscle contraction, weakness, and cramping following skeletal muscle stimulation. Failure in the mitochondrial genome of muscle cells in patients can be one of the reasons for the occurrence of this myotonia. Bioinformatics is an active research area aimed at developing intelligent systems for analyses of molecular biology. The purpose of this study was to investigate the structural changes of mitochondrial tRNA$^{lue(CUN)}$ in the presence of polymorphism A12308G with the help of bioinformatics tools. After PCR of mitochondrial tRNA$^{lue(CUN)}$ gene fragment from 28 Iranian patient's blood samples, all PCR products were subjected to *EcorI* enzyme and the result were detected by electrophoresis of 2.5% agarose gel (PCR-RFLP method). In three patients and one healthy person, A12308G polymorphism was observed as heteroplasmy. Statistical results did not show any significant difference (p-value>0.05) between this polymorphism and Non-Dystrophic Myotonia. To visualize the second structure of the mutated tRNA, we use the website mfold (unafold.rna.albany.edu/mfold) for predicting the secondary structure of RNA and DNA, mainly by using thermodynamic methods and RNAfold (rna.tbi.univie.ac.at/cgi bin/RNAWebSuite/RNAfold.cg) that predicts secondary structures of single stranded RNA or DNA sequences for partition function calculations and minimum free energy (MFE). Our results showed that A12308G nucleotide change alters the MFE from -15.00 kcal/mol to -14.80 kcal/mol thus has reduced the stability of the RNA molecule. Also, secondary structures of RNA show obvious changes which can show the importance of this polymorphism in the structure of the tRNA$^{lue(CUN)}$ molecule.

**Keywords:** Non-dystrophic Myotonia; A12308G Polymorphism; mtDNA, secondary structures.

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