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Mitochondrial transfer RNA mutations and hypertension

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ABSTRACT. Mutations in mitochondrial DNA have been found to be associated with hypertension. Of these, mitochondrial transfer RNA (mt-tRNA) is a hot spot for these pathogenic mutations. It is generally believed that these mutations may result in the failure of mt-tRNA metabolism, thereby worsening mitochondrial dysfunction and resulting in hypertension. mt-tRNA is known for its high frequency of polymorphisms and mutations, and the number of reports regarding mt-tRNA mutations and hypertension is increasing significantly. To better understand the molecular basis of maternally inherited hypertension, we reassessed the link between four mt-tRNA mutations (G15927A in $tRNA^{Thr}$, C7492T in $tRNA^{Ser(UCN)}$, A4386G in $tRNA^{Gln}$, and C14686T in $tRNA^{Glu}$) and hypertension. We first used the phylogenetic approach to investigate the deleterious roles of these mutations, then we used RNA Fold Web Server to predict the minimum free energy of these mt-tRNAs with and without mutations. Using the pathogenicity scoring system, we found that the G15927A and C7492T mutations are classified as pathogenic while all other studied mutations are neutral polymorphisms. Our study provides valuable information for the detection of pathogenic mt-tRNA mutations in hypertension.

Key words: Mitochondria; Mitochondrial tRNA mutations; Hypertension; Pathogenic mutations

