

عنوان مقاله:

The sequence variation of mitochondrial tRNA tyrosine and cysteine among Iranian women with idiopathic recurrent miscarriage: A case-control study

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خلاصه مقاله:

Background: Recurrent miscarriage is one of the most prevalent reproductive diseases. This phenomenon has several reasons, including maternal, hormonal, immunological, and parental genetic factors. Idiopathic recurrent miscarriage (IRM), with no distinctive etiology, involves about half of the recurrent miscarriage cases. Some mutations in mitochondrial DNA can lead to miscarriage. Mitochondrial tRNA (mt-tRNA) mutations cause nearly half of the mitochondrial disorders. **Objective:** To identify mt-tRNA Cys & Tyr gene mutations in Iranian women with IRM. **Materials and Methods:** In this case-control study, 100 Iranian women with IRM and 100 women as control without any history of miscarriage were investigated by polymerase chain reaction-single strand conformation polymorphism technique followed by gene sequencing. Bioinformatics analysis were done using human mitochondrial genome database, molecular evolutionary genetics analysis, mammalian mitochondrial-tRNA, etc. **Results:** Results showed 4 mt-tRNA mutations including 1 cysteine mt-tRNA mutation ($\Delta\text{A}24\text{C}>\text{T}$) and 3 tyrosine mt-tRNA mutations ($\Delta\text{A}68\text{T}>\text{A}$, $\Delta\text{A}49\text{C}>\text{T}$, and $\Delta\text{A}36\text{T}>\text{C}$) in our cases. **Conclusion:** Amongst the 4 mutations found, one was novel that is still not reported. Our bioinformatics analysis revealed that these mutations can be pathogenic. They occurred in tRNA-conserved regions and their secondary structure was changed, which can result in mitochondrial dysfunction. Mutations of these genes may help in the assessment of IRM. Further study of all 22 mt-tRNAs possible mutations is recommended to describe their etiologic role in IRM.

کلمات کلیدی:

Recurrent early pregnancy loss, mtDNA, SNPs, Heteroplasmy, سقط مکرر، DNA میتوکندری، SNP، هتروپلازمی.

