عنوان مقاله:

Novel Missense Mitochondrial ND۴L Gene Mutations in Friedreich's Ataxia

محل انتشار:

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خلاصه مقاله:

Objective(s) The mitochondrial defects in Friedreich's ataxia have been reported in many researches. Mitochondrial DNA is one of the candidates for defects in mitochondrion, and complex I is the first and one of the largest catalytic complexes of oxidative phosphorylation (OXPHOS) system. Materials and Methods We searched the mitochondrial ND۴L gene for mutations by TTGE and sequencing on ۳۰ FRDA patients and ۳۵ healthy controls. Results We found ۳ missense mutations [m.۱۰۵۰۶A>G (T۱۳A), m.۱۰۵۳۰G>A (V۲۱M), and m.۱۰۶۵۳G>A (A۶۲T)] in four patients whose m. ۱۰۵۳۰G>A and m.۱۰۶۵۳G>A were not reported previously. In two patients, heteroplasmic m.۱۰۵۳۰G>A mutation was detected. They showed a very early ataxia syndrome. Our results showed that the number of mutations in FRDA patients was higher than that in the control cases (P= ۰.۰۲۸۷). Conclusion Although this disease is due to nuclear gene mutation, the presence of these mutations might be responsible for further mitochondrial defects and the .increase of the gravity of the disease. Thus, it should be considered in patients with this disorder

کلمات کلیدی:

Friedreich's ataxia (FRDA), mtDNA, Mutation, ND۴L gene