عنوان مقاله:

(Identification of a Novel Mutation in Mitochondrial CO2 Gene in Patients with Familial Adenomatous Polyposis (FAP

محل انتشار:

چهارمین کنگره بین المللی سرطان های دستگاه گوارش (سال: 1397)

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خلاصه مقاله:

Introduction: Familial adenomatous polyposis (FAP) is an autosomal dominant predisposition to the development of polyposis in the colon and rectum. The first symptoms of FAP are diarrhea and blood in the stool. The incidence of the FAP disorder is one per 10000 newborns. The occurrence of FAP is associated with mutations in the APC tumor suppressor gene. The APC gene is located on chromosome 5q21 and is involved in cell proliferation control. Recent studies have shown that, in addition to nuclear genome, mitochondrial functional abnormalities are also as the most important carcinogenic factors in FAP. Nucleotide changes in the mitochondrial CO2 gene can be predisposing factors associated with FAP. Method: We screened mutations in the MT-CO2 gene in 26 patient with FAP disease and 20 healthy controls by PCR and sequencing. FAP symptoms in our patients diagnosed by specialists from Khatamolanbia Hospital of Tehran, Iran. Then, the analysis of pathogenicity of the MT-CO2 mutations was accomplished by the human mitochondrial genome database (Mitomap), PolyPhen, Sipred, Expacy and SIFT databases. Results: In 2 patients we found a heteroplasmic mutation 7602 A> C that cause change in amino acid (p. Q6P). This mutation is novel and has not previously been reported in any other disease. Conclusion: Based on our results of the pathogenicity prediction and bioinformatics assessments, this nucleotide change as missense mutation can be an important factor risk. The results of the mutation assay in the SIFT and Psipred databases, predict this mutation has affected on protein structure, and according to the result of ExPASy, hydrophobicity of mutant protein has reduced. PolyPhen results illustrates that this mutation is predicted to be probably damaging with a score of 0.999.

کلمات کلیدی:

.Familial adenomatous polyposis, Mitochondria, MT-CO2 gene, Mutation