عنوان مقاله:

Identification of an intronic mutation in CFTR in Iraqi children with cystic fibrosis

محل انتشار:

اولین کنفرانس ملی یافته های نوین زیست شناسی (سال: 1395)

تعداد صفحات اصل مقاله: 1

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خلاصه مقاله:

Cystic fibrosis (CF; MIM no. 219700) is the most common lethal genetic disease in the Caucasian population with an autosomal recessive inheritance. CF is caused by mutation in the cystic fibrosis transmembrane conductance regulator (CFTR) gene, located on the long arm of human chromosome 7 at the q31.2 locus, with 27 exons. In the present study we screened CFTR sequence in an Iraqi patient suffered from the classical form of cystic fibrosis.Peripheral blood samples were collected from patient and her parents. The 27 coding exons and flanking intronic regions of CFTR were amplified and sequenced. We found a homozygous variation; IVSII+12T> C in the enrolled patient. The parents were heterozygous for this variation. Bioinformatic analysis showed this variation has pathogenic effect and highly possibly causing CF. DNA analysis of the CFTR gene revealed that affected child has a known pathogenic mutation causing cryptic splice site,IVSII+12 T> C. This pathogenic variation should be concerned for the assessment of CF diagnosis, at risk -carrier detection and prenatal diagnosis.

کلمات کلیدی:

Cystic fibrosis, CFTR IVSII+12 T> C, the Iraqi patient