عنوان مقاله:

Identification of a novel single nucleotide polymorphism HADHA gene in an Iranian patient with Mitochondrial Trifunctional Protein Deficiency

محل انتشار:

دومین همایش ملی تازه های سلولی و مولکولی (سال: 1394)

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خلاصه مقاله:

Mitochondrial Trifunctional Protein deficiency is an autosomal recessive disorder due to the defect in the β-oxidation cycle of long-chain fatty acids. M-TFP deficiency are classified into 2 phenotypes: the more prevalent isolated LCHAD deficiency with defects of the α-subunits encoded by the HADHA (hydroxyacyl-CoA dehydrogenase α-subunit) gene and the less common pattern of complete M-TFP deficiencywith defectbothof HADHA or HADHB (hydroxyacyl-CoA .dehydrogenase ß-subunit) genes

کلمات کلیدی:

Mitochondrial Trifunctional Protein, HADHA