عنوان مقاله:
A genetic Assay of Three Patients in the Same Family with Holt-Oram Syndrome; a Case Report

محل انتشار:
مجله گزارش های بیوشیمی و زیست شناسی مولکولی, دوره 2, شماره 1 (سال: 1392)

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خلاصه مقاله:
Holt-Oram syndrome (HOS) is a developmental disorder inherited in an autosomal-dominant pattern. Affected organs are the heart and forelimbs with upper extremity skeletal defects and congenital heart malformation. In this study we present three cases of HOS in the same family. In one of these three individuals we detected a transition of C to T (CTG-GTT, V۲۰۵V) in exon ۷ of the TBX۵ gene. This nucleotide change causes no amino acid change and potential pathologic effects remain unknown.

کلمات کلیدی:
Congenital heart malformation, Holt-Oram syndrome, TBX۵ gene