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论著

一个中国先天性有汗性外胚层发育不良家系的GJB6基因筛查

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摘要:

目的: 先天性有汗性外胚层发育不良是一种常染色体显性遗传病,GJB6基因突变与之相关。调查一个来自中国的先天性有汗性外胚层发育不良家系与GJB6基因的关系。方法: 收集一个共有17个家系成员(患者8人,5男3女)的先天性有汗性外胚层发育不良家系,采集家系成员的外周血,抽提DNA。然后针对GJB6基因的每个外显子设计引物,应用聚合酶链式反应(PCR)方法扩增GJB6基因的整个编码区,测序、筛查突变。结果: 通过对家系患者的GJB6基因筛查,发现了一个杂合错义突变c.31G>A(p.G11R)。结论: GJB6基因错义突变c.31G>A(p.G11R)导致该家系先天性有汗性外胚层发育不良。

关键词: 毛发缺陷 先天性杵状指 错义突变 掌跖角化过度

Identification of a known GJB6 mutation in an autosomal dominant inherited Chinese family with hidrotic ectodermal dysplasia

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Abstract:

Objective: Mutation in the gap junction beta 6(GJB6)gene has been reported to be associated with an autosomal dominant disorder hidrotic ectodermal dysplasia(HED),characterized by congenital nail clubbing,alopecia and palmoplantar keratoderma.The aim of this study is to investigate relationship between genetic mutation in GJB6 and HED in an affected Chinese family. Methods: We selected a Chinese HED family consisting of a total of 17 individuals including 8 HED patients(5 males and 3 females).The whole coding region of GJB6 was amplified by polymerase chain reaction and sequenced. Results: Sequence analysis identified a heterozygous missense mutation c.31G>A(p.G11R)in GJB6 gene of affected individuals, but not in healthy individuals.Conclusion: A c.31G>A (p.G11R) missense mutation in GJB6 gene is the genotypic characteristic for HED in Chinese population.

Keywords: alopecia congenital nail clubbing missense mutation palmoplantar hyperkeratosis

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