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研究报告

两个弥漫性掌跖角化病家系的病理特征与基因突变分析

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摘要

为了明确两个弥漫性掌跖角化病家系的临床、病理特征、角蛋白9在局部组织中的表达情况及*KRT9*基因的突变，对2名先证者的手掌皮肤进行组织病理学、免疫组织化学分析，并用聚合酶链反应技术及直接测序分析的方法，对家系中46名成员的*KRT9*基因进行突变分析。发现两名先证者的表皮都呈显著的角化过度，颗粒层和棘层明显增厚，真皮浅层有轻度的炎症细胞浸润，上基底的棘层和颗粒层的角质形成细胞中都有特征性的空泡变性存在；角蛋白9只在棘层和颗粒层的角质形成细胞中特异性表达；两个家系患者分别存在*KRT9*基因的点突变N160S和L167S；说明这两个家系都属于表皮松解性掌跖角化病家系，*KRT9*基因N160S和L167S突变分别导致这两个家系发病。

关键词 表皮松解性掌跖角化病 角蛋白9 KRT9基因

分类号

Pathological features and gene mutation analysis in two pedigrees of diffuse palmoplantar keratoderma

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Abstract

<P>We report the clinical and pathological features of two pedigrees of palmoplantar keratoderma (PPK), the expression of keratin 9 (K9) in palm tissue and the mutation of the keratin 9 gene (<EM>KRT9</EM>). Histopathology and immunohistochemical assessment was performed to analyze the epidermis in the palm of the probands. Genomic DNA of 46 family individuals was used for amplification of exon 1 of <EM>KRT9</EM>. The mutations were determined by direct sequencing. Epidermal abnormalities in the palm of the two probands were characterized by vacuolar changes of suprabasal keratinocytes accom-panied by thickening of the living epidermis and stratum corneum. K9 was also expressed in particular epithelial tissues. Direct sequencing of polymerase chain reaction products revealed heterozygous missense mutations in exon 1 of <EM>KRT9</EM> (N160S and L167S) in the two families, respectively. N160S and L167S of <EM>KRT9</EM> are disease-causing mutations in these two Chinese pedigrees with epidermolytic palmoplanter keratoderma (EPPK).</P>

Key words epidermolytic palmoplanter keratoderma keratin 9 KRT9 gene

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