PRINT: ISSN 0972-3757 ONLINE: ISSN 2456-6330
International Journal of
HUMAN GENETICS

© IJHG 2021
PRINT: ISSN 0972-3757 ONLINE: ISSN 2456-6330
Int J Hum Genet, 21(4): 176-184 (2021)
DOI: 10.31901/24566330.2021/21.04.791

Identification of a Novel *KRT9* Frameshift Mutation in a Chinese Pedigree with Epidermolytic Palmoplantar Keratoderma

Haiou Jiang[1], Xiaoqing Zhao[2], Dan Yin[5], Song Wang[4], Hai Luo[1], Juan He[1], Jie Li[1], Linghan Gao[3] and Anli Shu[1,*]

[1]School of Basic Medicine, Hunan University of Medicine, Huaihua, China
[2]Department of Dermatology, Ruijin Hospital Affiliated to Shanghai Jiao Tong University School of Medicine, Shanghai, China
[3]Bio-X Institutes, Key Laboratory for the Genetics of Developmental and Neuropsychiatric Disorders, Ministry of Education, Shanghai Jiao Tong University, Shanghai, China
[4]Department of Dermatology, The First Affiliated Hospital, Hunan University of Medicine, Huaihua, China
[5]Health Center of Jinzhou Town, Ningxiang, China

KEYWORDS Deletion Mutation. Epidermolytic Palmoplantar Keratoderma (EPPK). Frameshift. Keratin. *KRT9*. Linkage Analysis

ABSTRACT Epidermolytic palmoplantar keratoderma (EPPK) is a genodermatosis with autosomal dominant inheritance model. It results from variants of keratin 9 (*KRT9*) or *KRT1* gene. In this study causative gene mapping in a Chinese EPPK family was performed with Two-point linkage analysis and haplotyping. Positive linkage results were obtained on 17q (Z_{max}=2.06, θ_{max}=0.0) at D17S799, which indicated *KRT9* to be the most responsible gene for the family. Subsequently, direct sequencing identified a novel frameshift mutation caused by a 5bp deletion (ΔGGAGG) in *KRT9* in all affected individuals but neither in the unaffected subjects nor in the 50 healthy unrelated controls. The frameshift changed the encoding of the following nine amino acids and resulted in a readthrough translation in exon 7. The data revealed that the novel frameshift mutation in *KRT9* was responsible for the Chinese EPPK pedigree. The researchers' findings broaden the spectrum of KRT9 variants and provide further evidence for the highly genetic heterogeneity of EPPK.