Hailey-Hailey disease: a novel mutation of the ATP2C1 gene in a Taiwanese family with divergent clinical presentation

蔡宗憲

Wang CC;Chao SC;Tsai TH

摘要

Abstract

Hailey-Hailey disease (HHD) is an autosomal-dominant hereditary disorder and heterozygous mutations in the ATP2C1 gene encoding a Ca~(2+) transport pump were mentioned in most of the cases.1-2 It is characterized by recurrent onset of vesicles and erosive, warty plaques predominantly over intertriginous areas, which can be initiated by various factors, such as friction, heat, perspiration and cutaneous infections. We identified a novel mutation of the ATP2C1 gene in a Taiwanese family with divergent clinical presentation.A 50-year-old man, wheelchair-bound by poliomyelitis, presented with generalized reddish maceration and plaques over the scalp, trunk, bilateral axillary and inguinal areas which have waxed and waned for more than 5 years (fig. 1). Histopathology revealed supra-basal separation with individual acantholytic cells, which was consistent with HHD. Mutation analysis of ATP2C1 gene from the peripheral blood of the patient and his also-affectedmother were conducted by polymerase chain reaction (PCR) amplification and then analysed on an ABI 377/ automatic sequencer (Advanced Biotechnologies, Columbia, MD, USA).3 The result indicated a novel mutation site in both cases - 28-base-pair deletion between exon 17 and intron-exon boundary of ATP2C1 gene (either GGACAGACCAGAGATTTGTTTTATGAAA at nudeotide 1593-1620 position or GACAGACCAGAGATTTGTTT-TATGAAAG at nucleotide 1594-1621 position) - which resulted in frameshift and premature termination codon.