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

*Editor*

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-affected mother 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 nucleotide 1593–1620 position or GACAGACCAGAGATTTGTTT-TATGAAAG at nucleotide 1594–1621 position) – which resulted in frameshift and premature termination codon.

His initial treatment had been with short-term, low dose prednisolone (0.15–0.3 mg/kg/day) to control this problem. Despite vigorous therapeutic trials, including oral acitretin, oral and topical antibiotics (tetracycline, bacitracin-neomycin ointment), topical calcitriol, intralesional botulinium toxin type A injection[4] and topical gentamicin[5] soaking, frequent superinfections and near constant superficial erosions accompanied by serous exudates were noted over his buttock, groin and axillary areas. Within all treatment modalities, the patient showed systemic steroid dependence. In contrast, the skin eruptions of his mother were, if present, limited to the axilla, and she has been lesion free for more than two years. Therefore, we proposed that frequent friction over the body folds in our wheelchair-bound patient may have led to such a recalcitrant course, given the divergent severity of presentation in the same family. The above circumstance is compatible with the observation of Dobson-Stone *et al.* that modifying genes and/or environmental factors may greatly influence the clinical presentation of HHD.[6]

Our findings suggest that the novel mutation site identified in the *ATP2C1* gene and local factors encountered (e.g. friction and heat) may both play important roles in the pathogenesis of HHD.

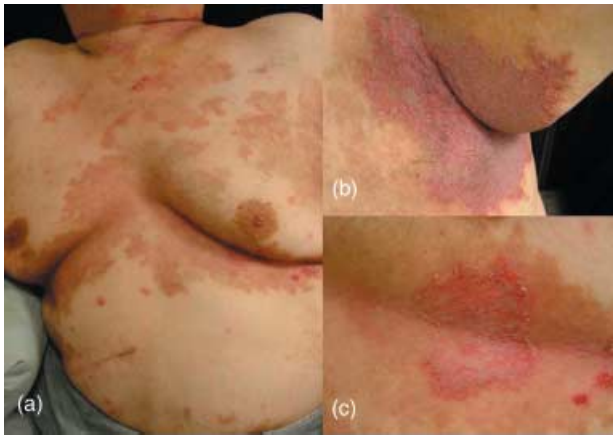


**fig. 1** Multiple well demarcated, erythematous, erosive plaques with vegetative change over (a) anterior chest (b) bilateral axillary (c) infra-mammary and groin area.

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# Zygomatic flap combined with a palatal mucosal graft in the reconstruction of eyelid defects

*Editor*

Full-thickness defect of more than 30% of the width of the eyelid cannot be closed directly, and the skin as well as the conjunctiva have to be reconstructed; therefore, various techniques to repair large full-thickness defects have been reported.

A 76-year-old woman presented with a squamous cell carcinoma on the left periocular region involving the upper and lower eyelid (fig. 1a). Surgical excision was performed with resection of the external one-fourth of the upper eyelid and the entire lower eyelid (fig. 2). A hard palate mucosa graft was taken and inserted to create the upper and lower lid conjunctiva. The both eyelid were reconstructed with a zygomatic flap. The functional and cosmetic results were very acceptable (fig. 1b) without complications like ectropion, entropion or ptosis. The donor site healed satisfactorily. The patient remains free of the tumour in a 6-year follow-up.

Full-thickness reconstruction requires the use of additional material for the conjunctival lining.[1] One of the most important points in the eyelid reconstruction is the construction of a supporting system to substitute for the tarsus. The hard palate mucosa is thick and rigid compared with the oral mucosa and thus provides protection and support simultaneously.[2]

The advantages of a palatal mucosa for conjunctival reconstruction include rigidity, adequate curvature, good

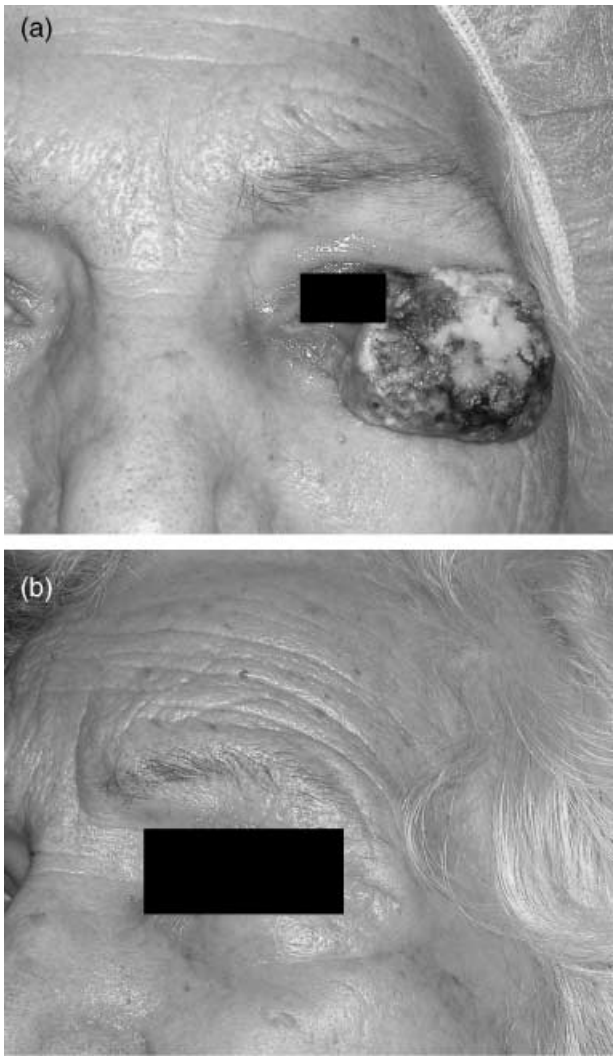


**fig. 1** Squamous cell carcinoma of the left eye (a) and 3 months after surgery (b).