GENETICS AND GENODERMATOSES

A CASE OF A 56-YEAR OLD MALE WITH HAILEY-HAILEY DISEASE: A PROBLEM-BASED APPROACH

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Background: Hailey-Hailey disease (HHD), is a rare autosomal dominant disorder affecting adhesion of epidermal keratinocytes. Among the eleven society-accredited institutions, only 24 cases have been reported in the past seven years. With onset from puberty to the fourth decade, this intraepidermal blistering disorder is characterized by painful blistering, erosions, maceration, and frequent secondary infection in the flexural areas. HHD is a chronic condition with multiple recurrences and limited therapeutic options. We present an atypical case that shows promising response to humectants and keratolytics in addition to first-line treatment.

Observation: A 56-year-old male working in field work construction for 26 years presented with history of pruritic, hyperpigmented plaques on intertriginous areas accompanied by small, erythematous crops of papulovesicles on the flexural folds that were pruritic and symmetric. These were triggered and exacerbated by the heat, friction and sweating. The lesions were transient and will resolve when moved to a cool environment. There is a remitting and relapsing course, with similar lesions in his 52-year old brother. A punch biopsy showed findings consistent with HHD. After disseminated dermatophytosis was addressed with oral antifungals, the patient was managed with topical betamethasone plus gentamycin. Chlorhexedine gluconate 4% wash was used, however stopped due to the development of cutaneous hypersensitivity. Lastly, a humectant with lactic plus salicylic acid was used with above average results.

Key Message: Levels of other ATPase proteins may influence expressivity of HHD that could account for atypical presentations, in contrast to patients only carrying an ATP2C1 mutation. This case supports aminoglycoside antibiotics' potential to reverse the effects of pathogenic nonsense mutations in this human genetic disorder. A humectant-keratolytic topical combination appears beneficial for mild disease and shows promise for future therapy in this genodermatosis that is known to having limited therapeutic options.