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PhotoDermDiagnosis

Exudative, macerated, fissured flexures in a young female

Nabeela Shahzadi, Faria Asad, Zahida Rani

Department of Dermatology, Unit I, King Edward Medical University/ Mayo Hospital, Lahore

Introduction

A 26-year-old female presented with 3-year history of recurrent scaly, exudative, erythematous lesions involving sides of neck, axillae, inframammary region and groins. She developed small superficial vesicles that ruptured spontaneously or on scratching. These coalesced to form macerated plaques. She had symptoms of pain, discomfort and itching. Her lesions flared up every summer to remit partially in winters. The disease responded partially to topical steroids. Her mother and elder sister had similar eruption but of milder intensity.

On examination, malodorous, moist, macerated, erythematous plaques were present on sides of her neck, axillae (**Figure 1**), antecubital fossae, inframammary region and groin in a bilateral and symmetrical fashion. Histopathology from inframammary region is shown in **Figure 2** and **3**.

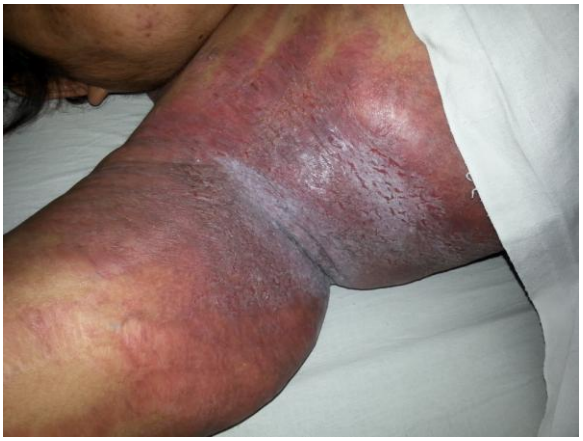

Figure 1

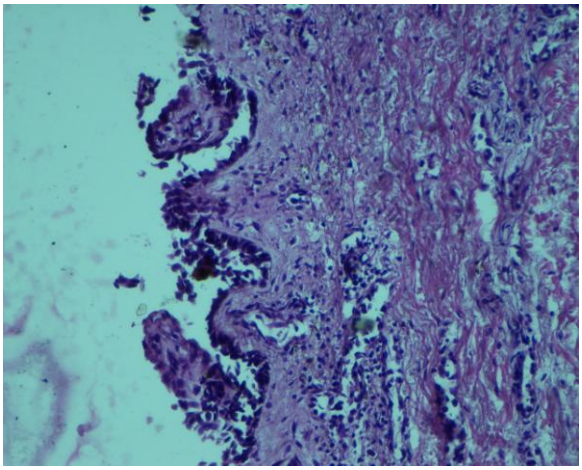

Figure 2

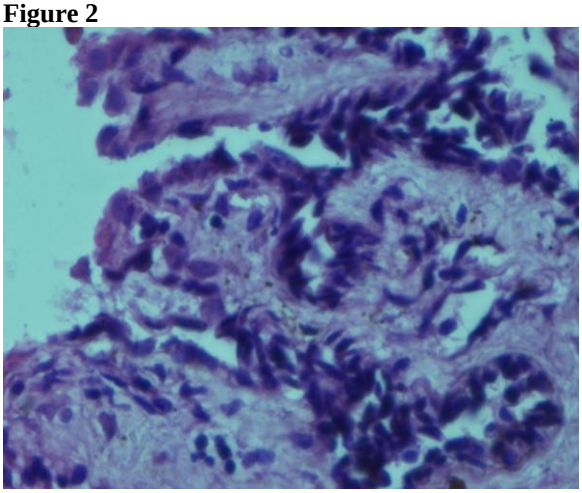

Figure 3

What is the diagnosis?

Address for correspondence
Dr. Nabeela Shahzadi,
Department of Dermatology, Unit I,
King Edward Medical University/
Mayo Hospital, Lahore
Email: ibrahimsohaib20@gmail.com

Diagnosis

Hailey-Hailey disease (HHD)

Histopathological examination showed suprabasal partial acantholysis. "Dilapidated brick wall" appearance was also seen. In the dermis, lymphocytic infiltrate was noted at places.

Discussion

HHD is characterized by abnormal desmosomal adhesion between keratinocytes. ATP2C1 has been identified as the causative genes for Hailey–Hailey disease which encodes a secretory pathway Ca2+/Mn2+-ATPase (SPCA1) found in the Golgi apparatus. Mutations in this gene cause the disease by altering the formation or stability of desmosomes.[1]

HHD is a rare disorder. Its prevalence is unknown, since many patients are not diagnosed accurately or never seek treatment. The age of onset and clinical manifestations of HHD vary widely within families.[2] In our case, the patient had the classical spectrum of the disease and pursued remitting and relapsing course. Her elder sister and mother had such a mild manifestation that they never sought advice from a tertiary care set up.

HHD is inherited in an autosomal dominant manner with complete penetrance and variable expressivity. Only two-thirds of patients have a family history of HHD, as in our case showing strongly positive family history. Absence of family history could be explained by de novo mutations or lack of phenotypic expression in affected family members. Certain environmental and/or other genetic factors might also add up to the diversity of interfamily clinical presentation.

The onset of disease typically occurs in the second and third decades of life. Our patient had her disease started since the age of 25 years. Clinical manifestations primarily involve the flexural areas in a symmetric fashion (groin, axillae, lateral neck, submammary region, and perineum). Flaccid vesicles on erythematous to normal skin are the first manifestation and often pass unnoticed as they are superficial and of short duration. Large, macerated, exudative plaques of superficial erosions with crusting are usually seen at the time of the presentation. Further progression to large vegetative malodorous plaques with painful fissures can occur. Flexural disease may be disabling, especially if the groin is involved. Longitudinal white bands of the nails have been described in about 70 percent of patients with HHD and can be a clue to the diagnosis. Mucosal involvement is rare.[3]

The triggering factors like friction, heat, sweating, constrictive clothing, physical trauma, stress and menstruation have been described.[4] Our patient also had exaggeration of symptoms during sweating, friction and heat.

Histopathologic examination reveals widespread acantholysis resulting in suprabasal clefts, vesicles and bullae. Areas of partial acantholysis form layers of intraepidermal and suprabasal detached keratinocytes with the appearance of a "dilapidated brick wall". Dyskeratosis is usually mild. The papillary dermis contains a sparse perivascular lymphocytic infiltrate with scattered eosinophils.[5]

Treatment can be topical in the form of long-term antibiotics and/or antifungals and topical corticosteroids which may be used intermittently, as an adjunct to topical antibiotics and antifungals. Tacrolimus 0.1% ointment, alone or in combination with other treatments,

has been reported as beneficial.[6] In addition, botulinum toxin injection, in affected areas, has been used as an adjunctive treatment to reduce sweating and maceration.[7]

Systemic therapeutic options include cyclosporin 2.5 to 3.5 mg/kg per day[8] and acitretin 25 mg/day.[9] Surgical and destructive methods, including carbon dioxide laser or 595 nm pulsed dye laser ablation,[10] cold atmospheric plasma,[11] dermabrasion[12] and photodynamic therapy with 5-aminolevulinic acid,[13] have been used in patients with recalcitrant disease.

The long-term benefits of medical and/or surgical approaches are uncertain and recurrence is common. Patients with HHD have a normal life expectancy, although with significantly impaired quality of life.

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