

Case Number 8

Hailey- Hailey Disease

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Case summary:

Demographic details:

Mr. J.M., Male, Fgura, 73 years of age.

Mr. J.M. is a 73-year old gentleman who was diagnosed with Hailey-Hailey disease at the age of 28 and is being followed up at Dermatology out-patients. He has suffered since early adult life from recurrent maceration of the skin especially at the groin, inner thighs, axillae and neck folds. These areas are accompanied by erythema, itching, pain and oozing of a malodorous fluid and are waxing and waning in nature. Currently the patient has 2 areas of skin maceration over both inner thighs, worse on the left side with involvement of the left scrotum.

Presenting Complaint:

Pain and irritation of the skin of the left inner thigh and scrotum with maceration.

History of Presenting Complaint:

A 73-year old gentleman with a 45 year history of Hailey-Hailey disease seen as an out-patient at Sir Paul Boffa Hospital. When the patient was in his late twenties, he noticed several areas of itching followed by redness and pain in varying locations such as inner thighs, groin, axillae and neck folds. The condition was waxing and waning in nature. At this point it was not severe enough to warrant medical attention. During the following 5-6 years, the development of new affected areas was becoming more severe with resulting fissuring, severe pain and oozing of a malodorous fluid. This prompted the patient to seek medical attention and a clinical diagnosis of Hailey- Hailey disease was made which was subsequently confirmed with a skin biopsy.

The patient describes a phase of itching 2 days prior to development of redness, fissuring and oozing of fluid which is occasionally accompanied by pus. The resulting pain interferes with walking especially when the inner thighs and inguino-scrotal folds are affected. The condition is worse in summer, during periods of stress, when wearing nylon clothing and following laborious work. Relieving factors include swimming, increased frequency of bathing and application of a mild topical steroid cream.

Currently the patient has an affected area over the left inner thigh and scrotum. This episode was described by the patient as being mild when compared to previous episodes, but the patient would like to start treatment to prevent further maceration of the skin.

Past medical and surgical history:

Past medical history

Hypertension

Diabetes Mellitus Type 2

Asthma

Ischaemic heart disease with previous episodes of Angina (Angiogram and PCI done)

Past surgical history

Endoscopy for epigastric pain done 30 years and 20 years ago; both were negative.

Angiogram and PCI done 7 years ago.

Drug history:

Drug	Dosage	Frequency	Type	Reason
Metformin	500mg	TDS	Biguanide	Type 2 Diabetes Mellitus
Perindopril	8mg	BD	ACE Inhibitor	Hypertension
Amlodipine	5mg	Once daily	Calcium Channel Blocker	Hypertension
Aspirin	75mg	Once daily	Anti-platelet	Ischaemic Heart disease
Simvastatin	40mg	Nocte	Statins	Ischaemic Heart disease
Salbutamol	-	PRN	Bronchodilator	Asthma

Allergies:

Betamethasone Cream

Family history:

J.M.'s mother suffered from sub-mammary fold fissuring, Hailey-Hailey disease, however no histological diagnosis was made.

J.M.'s sister was diagnosed with Hailey-Hailey disease a few years after his diagnosis.

J.M. has 2 sons and a daughter. One of the sons suffers from eczema and Hailey-Hailey was excluded following investigations. The youngest son is suspected to be suffering from the same condition but has not yet sought medical attention.

Social History:

Occupation: The patient is a retired civil servant.

Status: J.M. is a widower, since 10 years ago. The condition did not affect the relationship with his wife, since a physician had put their mind at rest that it was not contagious.

Habits: The patient is an ex-smoker since the age of 15 years but stopped 25 years ago. He smoked 2 packets daily. He consumes alcohol socially.

Lifestyle: The patient led a very busy lifestyle especially due to his occupation. The condition affected him both physically and psychologically, due to the pain, odour, and persistent fear of having a sexually transmitted disease prior to the correct diagnosis.

Systemic inquiry:

- General Health: Patient looks well in general. No loss of appetite or weight
- Cardiovascular System: None
- Respiratory System: Recent onset of shortness of breath on activity especially when climbing stairs and slopes. No accompanying chest pain, cough or lower limb oedema.
- Gastrointestinal System: None
- Genitourinary System: None
- Central Nervous System: None
- Musculoskeletal System: None
- Endocrine System: None

Discussion of results of general and specific examinations:

On general inspection the patient appeared well and not distressed. On examination of the affected areas, areas of hyperpigmentation were noted over both axillae, with the left axilla being more prominent, and both inner thighs. There was erythema, maceration and fissuring over the left inner thigh extending to the left scrotum (Figure 1). There were no vesicles or discharge however a slight malodour was noted.

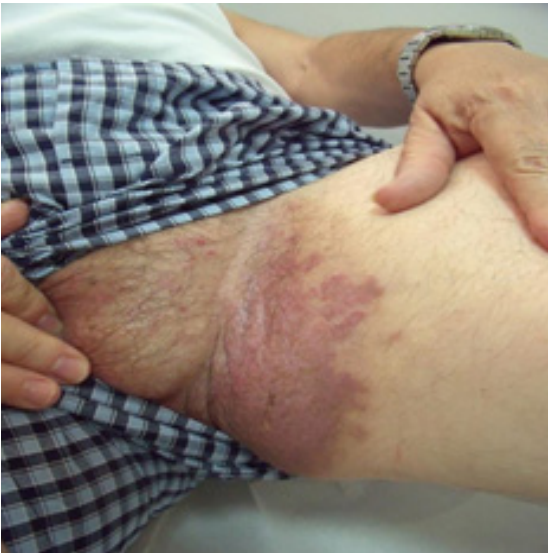


Figure 1: The affected area at presentation over the left inner thigh/scrotum.

Areas of hyperpigmentation and dryness were also noted over both shins, likely due to venous insufficiency. Mild lower limb pitting oedema was also present.

Differential diagnosis of intertrigo:

- Seborrhoeic Eczema
- Flexural Psoriasis
- Candidiasis
- Tinea Corporis
- Erythrasma
- Acanthosis Nigricans
- Pemphigus Vegetans

Diagnostic procedures:

Laboratory Exams:

Skin Biopsy: Date: 20/5/2000

Test: 5mm punch biopsy from right axilla.

Justification for test: To confirm diagnosis.

Result: Sections from the skin show epidermal hyperplasia with suprabasal acantholysis that involves the spinous layer giving a 'dilapidated brick wall' appearance. Corps ronds and grains are infrequent.

Conclusion: Hailey-Hailey disease.

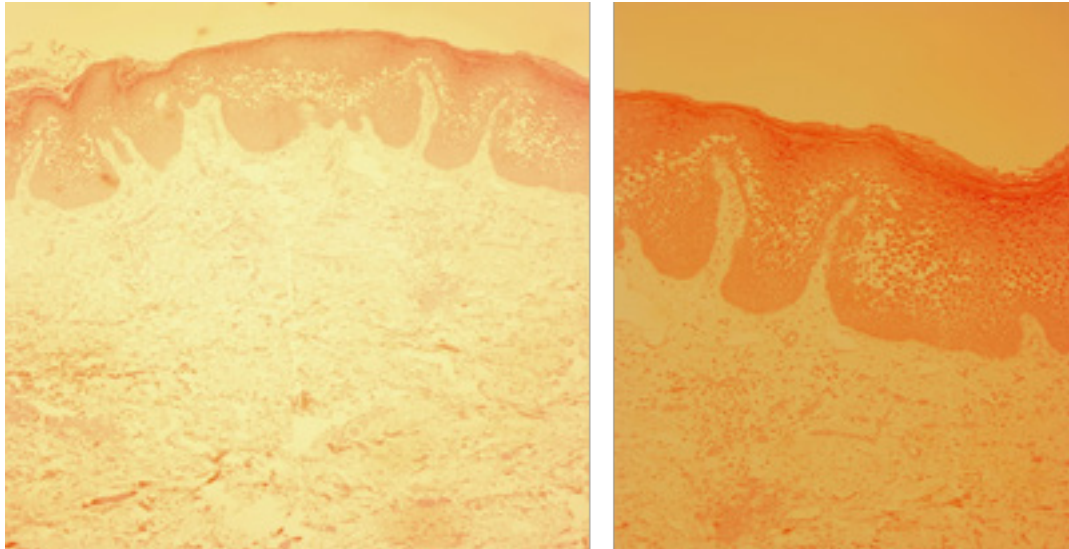


Figure 2: Histology photos of Mr. J.M. showing characteristic features of Hailey-Hailey disease, including epidermal hyperplasia with suprabasal acantholysis.

Diagnosis:

Hailey-Hailey disease is a rare autosomal dominant blistering disorder of the skin which results in defective adhesion of keratinocytes resulting in breakdown of affected skin layers¹. First signs of the condition usually appear between ages of 15 to 40, 28 years in the case of Mr. J.M. Affected individuals complain of red, scaly areas or small blisters at areas of friction such as inner thighs and axillae, as in this patient. These areas may become secondarily infected by bacteria, fungi or viruses explaining the malodorous discharge the patient describes². The clinical presentation and strong family history pertaining to Mr. J.M. give a strong indication of Hailey-Hailey disease, however a biopsy was still done to confirm diagnosis³. The skin biopsy showed suprabasal acantholysis involving the spinous layer giving a 'dilapidated brick wall' appearance, which is typical of Hailey-Hailey disease⁶.

Final treatment and follow ups:

Patient was advised to apply a 1:1 mixture of Hydrocortisone butyrate 0.1% cream and Clotrimazole Cream to affected areas until resolution.

In addition, he was prescribed Ciprofloxacin 500mg BD PO x 2weeks.

Fact Box 8:

Title: Hailey-Hailey disease

General overview: Hailey-Hailey disease is a rare autosomal dominant blistering condition which was first described by the Hailey brothers in 1939. It is sometimes called ‘familial benign chronic pemphigus’, but this creates confusion in the medical literature. Pemphigus is a general term for a group of autoimmune blistering skin disorders, whilst Hailey-Hailey is not an autoimmune disorder but a distinct genetic disorder⁴. It can occur at any age, but usually appears in the third or fourth decade. Clinically the condition has a fluctuating course and both the activity and the affected areas may vary⁵.

The defect responsible has been identified on ATP2C1 gene found on chromosome 3q21-24. This gene codes for the protein Secretory Pathway Calcium/manganese-ATPase (SPCA1), which is a calcium and manganese pump. The keratinocytes normally stick together via desmosomes and due to this defect leading to insufficient calcium, the desmosomes do not assemble properly^{2,6-7}. Normally the cells are packed together tightly like bricks and mortar. Patients with this Hailey-Hailey disease have defective ‘mortar’ leading to the cells falling apart, reminiscent of a dilapidated brick wall⁶.

Signs and Symptoms:

Clinically it typically presents with a painful erosive rash in the intertriginous areas namely the axillary (Figure 3), inguinal, perineal (Figure 1) and neck folds. At first they come and go leaving no scars. They can become thickened if present for some time. The skin often breaks down leaving painful fissures. Secondary bacterial infection gives rise to a malodour^{2,6}. Mucous membranes are less affected. Longitudinal white lines on the nails may be found.



Figure 3: An affected area at the axilla during a previous flare up.

Epidemiology: No precise data is available on the incidence of Hailey-Hailey disease. Many patients lack an accurate diagnosis or do not seek treatment. It causes discomfort but is not a life threatening condition and does not affect life expectancy. Both sexes are affected equally and there is no apparent difference in prevalence among different ethnic groups⁷.

Aggravating factors: Mechanical irritation, hyperhidrosis, bacterial, fungal and viral super infection and maceration of the skin are considered important aggravating factors⁸. Most patients find that their condition worsens during the summer months⁶.

Prognosis: Many patients have long remissions and may improve with age.

Management: There is no cure for the disease and treatment aims at reducing symptoms and preventing flare ups. The mainstay of treatment includes topical steroids and appropriate use of topical or systemic antimicrobials. It is important to educate patients to avoid aggravating factors such as sweating, synthetic and tight clothing and being overweight. Emphasis should be made on strict glycaemic control in diabetics. Retinoids, ciclosporin, dapsone, methotrexate, botulinum toxin to reduce sweating and laser therapy have been used with variable results⁶.

References:

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