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Case Study

HAILEY-HAILEY DISEASE – A CASE REPORT

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ABSTRACT

Hailey-Hailey disease (HHD) is a rare autosomal dominant skin condition discovered in 1939 by Hailey brothers. Prevalence is found to be around 1 in 50,000 and is relatively uncommon in India. First onset of disease occurs between 20 to 40 years of age, usually presented in the 3rd and 4th decades of life. Here, we report a case of a 50-year-old female who presented with clinical features of the HHD and showed greater response to the treatment.

Keywords: Familial pemphigus, Hailey-Hailey, ATP2C1 gene, Acantholysis, Dilapidated brick wall.

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INTRODUCTION

Hailey-Hailey disease (HHD) also called benign familial pemphigus is a rare autosomal dominant disorder that affects the adhesion of epidermal keratinocytes which recurs with remitting and relapsing episodes. It is caused by mutations in the ATP2C1 gene which results in altered cellular connections within desmosomes and adherens junctions of epidermis [1]. HHD is primarily localized in intertriginous areas usually characterized by painful blistering, erosions, maceration, and frequent secondary infection in flexural areas such as neck, axillae, groins, and perineum [1,2].

CASE REPORT

A 50-year-old married female with h/o T2DM and microadenoma was presented to the medical OP department with itchy, erythematous papules mainly over the flexural areas on and off since 20 years (Fig. 1). She got temporary relief from symptoms by taking homeopathic treatments and also allopathic treatments from outside. The lesions reappeared in the intertriginous areas. Skin biopsy obtained from erythematous plaques on the chest region revealed irregular acanthosis with a diminished or granular layer, a subcorneal pustule composed primarily of neutrophils and occasional eosinophils, and acantholytic keratinocytes. All routine hematological tests were within normal limits. Based on the clinical and histological findings, it was diagnosed as HHD. The patient had a positive family history (her father and nephew) for this disease as well.

Initially, the patient was treated with immunomodulators and topical antibiotics T. dapsone 100 mg OD and sodium fusidate cream, having transient remission in the 1st year of diagnosis. The skin lesions reappeared in 2018 and were treated with systemic corticosteroids. The patient developed increased blood sugar levels associated with the use of steroids. She developed wide spread staphylococcal skin infection with rapid generalization of the lesions. Inj. clindamycin 600 mg BD improved the skin condition. New lesion developed on foot and managed with prednisolone 40 mg OD and T clindamycin 300 mg BD daily.

DISCUSSION

The incidence of HHD is 1 in 50,000 and is relatively uncommon in India. About two-third (60%) of patients have a positive family history. The disorder becomes apparent after puberty, usually by the third or fourth decade but symptoms can develop at any age [2]. The initial lesion may be red, scaly area, or a fluid-filled blister which ruptures easily and becomes macerated or crusted. The lesions may develop a yellow crusty overlying layer. In many cases, the rash may be itchy or cause a burning sensation. The lesions can separate leaving painful, cracked skin. Secondary infection of the skin lesions can occur and may cause an unpleasant odor. Sunlight, heat, sweating, and friction often aggravate the disorder [3,4].

HHD is caused by mutations in the ATP2C1 gene at 3q22.1, which encodes the ATP powered, magnesium-dependent calcium pump protein hSPCA1, its function is to maintain normal intracellular concentration of free calcium (Ca^2+) by sequestering Ca^2+ into the Golgi apparatus. Calcium homeostasis in keratinocytes is involved in epidermal differentiation, barrier repair, cell-cell adhesion, and keratinocyte motility. Genetic defect results in altered cellular connections within desmosomes and adherens junctions of epidermis, secondary to high cytosolic calcium levels. Other manifestations include oxidative stress and microRNAs [4,5].

Histopathology has a key role in diagnosis of HHD with characteristic dilapidated brick wall appearance, intracellular immunoglobulin G is not detected in epidermis by direct immunofluorescence staining in contrast to autoimmune pemphigus [5]. Treatment of HHD is very challenging; there is no specific therapy for HHD. Its management involves control of exacerbating factors, secondary infections, and

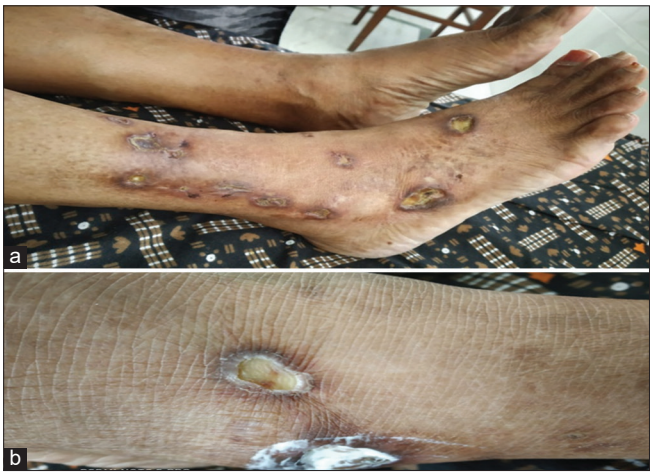

Fig. 1: (a and b) Erythematous papules over the flexure areas

cutaneous inflammation. Topical therapy includes antibiotics, steroids, tacrolimus, and calcitriol, whereas systemic therapy includes antibiotics (clindamycin, gentamicin, and mupirocin), steroids, methotrexate, tacrolimus, dapsone, and thalidomide [3-5].

CONCLUSION

The novelty of our report is that disease was diagnosed in 2018 after 20 years of history during the sixth decade. Delay in the diagnosis eventually leads to worsening of symptoms. High-dose steroid was not given to this patient as she was a diabetic patient so clindamycin was administered systemically and sodium fusidate topically. The patient responded well to the treatment and got discharged from the hospital.

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AUTHORS' CONTRIBUTIONS

Dr. Sunu Kurian treating doctor provided the clinical details of the case and reviewed the manuscript, Ms. Anjaly P Nair involved in the preparation, reviewing, and editing of the manuscript, and Dr. Siby Joseph and Ms. Lakshmi R were involved in the organizing, reviewing, and editing of the manuscript.

CONFLICTS OF INTEREST

The authors declare no conflicts of interest.

AUTHORS' FUNDING

Nil.

ETHICAL STATEMENT

Ethical approval is not applicable for case report in our institution.

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