

**GENITAL HAILEY - HAILEY DISEASE: A CASE REPORT**

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None

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**Abstract**

Hailey - Hailey disease is a rare autosomal dominant acantholytic disorder, previously not reported from Nepal. We report a case of 30 years old female who presented with pruritic hyperkeratotic papules and plaques on vulva, perianal area and inner left thigh for a period of one year. Biopsy from the lesion showed suprabasal acantholysis with loss of intercellular bridges resulting in a dilapidated brick-wall appearance; characteristic of Hailey - Hailey disease. Treatment of this disease till date is far from satisfactory.

**Key words:** acantholysis; Hailey - Hailey disease; Nepal

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**Introduction**

Hailey-Hailey disease is a rare autosomal dominant acantholytic disorder. It is characterized clinically by a recurrent eruption of vesicles and bullae at the sites of friction and intertriginous areas. Histopathology is diagnostic of Hailey-Hailey disease. We present a case with an atypical presentation involving vulva, previously not reported from Nepal.

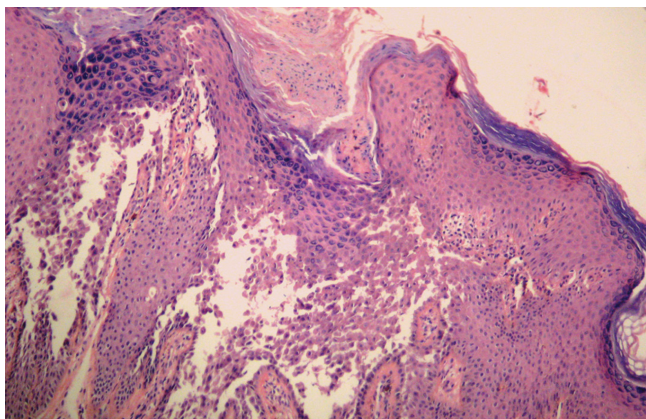
**Case Report**

A 30 year old female presented in the Department of Dermatology, Nepal Medical College and Teaching Hospital, with pruritic hyperkeratotic plaques on vulva, perianal area and inner left thigh for a period of one year. She initially had itching on the vulvar area. A month later, she noticed hyperkeratotic small raised lesion in vulva, which over three months, coalesced to form bigger lesions and spread to perianal area and inner left thigh. She experienced increase itching during friction and sweating but it did not aggravate during menstruation or stress. She denied history of similar disease in her family. She neither took medical advice nor medication prior to coming to our department. Local examination revealed violaceous to brownish irregular hyperkeratotic papules and plaques of different sizes, with well defined margin on lower one third of bilateral labia majora which extended to involve perianal area. There was also a plaque near medial aspect of inner left thigh near groin area (Fig. 1). Systemic examination showed no other abnormality. Biopsy revealed characteristic

features of Hailey-Hailey disease (Fig. 2) showing large separation of detached stratum malpighii cells with loss of their intercellular bridge (acantholysis effect) in suprabasal portions. Detached epidermis showed dilapidated brick wall appearance, which was consistent with Hailey-Hailey disease. Immunofluorescence test, due to unavailability, could not be done in our set up. The patient was treated with oral Doxycycline and topical clobetasol propionate 0.05% cream and Tacrolimus 0.1% ointment. A month later follow up examination revealed marked clinical improvement. Patient was then continued with topical tacrolimus.



**Figure 1. Multiple hyperkeratotic papules and plaque on vulva and near groin area**



**Figure 2. Detached epidermis showing characteristic dilapidated brick wall appearance**

## Discussion

Hailey-Hailey disease also known as Familial benign chronic pemphigus, was first described in 1939. It is an autosomal dominant acantholytic disorder which clinically presents as recurrent painful or pruritic fragile, vesicles and erosions in intertriginous areas involving axillary folds, groin, submammary region, and neck folds [1]. Patients mostly present with symptoms during the second or third decade of life and suffer from chronic, relapsing outbreaks [2]. Our patient presented with an atypical presentation involving vulva with hyperkeratotic plaques rather than the characteristic vesicle or erosion. Literature review quotes presence of lesion in atypical sites like with symmetrical distribution limited to the upper chest and anterior aspects of the upper arms and neck [3], erythroderma [4], conjunctivae [5] or mucosae [6,7]. The triggering factors like friction, heat, sweating, constrictive clothing, physical trauma, stress and menstruation have been attributed. Our patient also had exaggeration of symptoms during sweating and friction. Characteristic histopathological examination shows widespread suprabasal acantholysis with loss of intercellular bridges, which results in a dilapidated brick-wall appearance and similar picture was also seen in our patient. Recently studies have shown that Hailey-Hailey disease occurs due to the result of mutations in the ATP2C1 gene, which encodes  $\text{Ca}^{2+}/\text{Mn}^{2+}$ -ATPase protein 1 (hSPCA1), which is localized to the Golgi apparatus [1]. Keratinocytes which shows ATP2C1 mutation, have deficient  $\text{Ca}^{2+}$ -signaling, with dysregulated sorting and glycosylation of desmosomal proteins, giving rise to epidermal defects in skin lesions. Patients with Hailey-Hailey disease, a total of 98 ATP2C1 mutations have been reported worldwide. Linkage analysis has localized the gene locus to chromosome 3q21-q24 [8]. Colonization and secondary infections with bacterial, fungal, or viral microorganisms are known to be associated with Hailey-Hailey disease. Squamous-cell carcinoma that is rare can also occur [9]. The frequency of exacerbations may be decreased by wearing light weight clothing and avoiding activities that result in sweating or skin friction. Treatment option includes topical antimicrobials, steroids

and intralesional steroids. Systemic therapy includes oral antimicrobials and few case reports describe use of cyclosporin, acitretin and methotrexate. Surgical methods like dermabrasion, CO<sub>2</sub> or erbium-YAG laser vaporization and others like 5-aminolevulinic acid photodynamic therapy, botulinum toxin have been used with success. Surgical management with wide local excision of affected skin folds has a high complication rate. Refractory Hailey-Hailey disease may benefit from local electron-beam therapy [3,11-14].

## Conclusion

This is very rare disease and no case has been reported from Nepal till date. Histopathology is an important diagnostic tool to diagnose Hailey-Hailey disease. Due to its relapsing and remitting course of the disease there is need to have effective treatment options in future to improve quality of life of the patient with Hailey-Hailey disease.

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