Manipal Journal of Nursing and Health Sciences

Volume 6 | Issue 1 Article 22

1-1-2020

A rare case: Hailey-Hailey disease

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Case Report

A rare case: Hailey-Hailey disease

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Abstract

The Hailey-Hailey disease is a rare autosomal dominant disorder triggered by a genetic mutation that is characterized by the growth of repeated blisters and erosions in the intertriginous areas. Commonly the treatment is focused on the symptoms and severity of the condition. Currently, there is no specific treatment available for this disorder.

Keywords: Autosomal disorder, Gene Mutation, Hailey-Hailey disease

Introduction

Hailey-Hailey disease (HHD) is a rare disorder. Due to the lack of accurate diagnosis and many patients not seeking treatment, there is no data available on the prevalence rate. Hailey-Hailey disease is a genetic skin disease induced by mutations in the ATP2C1 gene. This disease is hereditary in an autosomal dominant manner. The signs and symptoms of this rare disorder include blisters and sore rash in skin folds such as the neck, armpits, under the breasts, groin, and in between the buttocks. Symptoms are seen in the months of summer due to heat, sweating (excessive perspiration), and friction (NORD, 2017). HHD is difficult to treat, even with combinations of topical and systemic antibiotic and immunomodulatory medications (Fernandez, 2017). Thus, HHD causes significant morbidity and adverse effects on quality of life (QOL) for affected patients.

Case report

During a home visit, a 53 years old married woman had complaints of persistent itchy, recurring painful lesions filled with pus on the abdomen, for 10 years. The lesions were oozing with foul-smelling discharge most of the time. These lesions intensified on exposure to the sun during the summer. The first episode was ten years back when she suddenly noticed the appearance of 4 – 5 itchy vesicles and also suffered from pain and watery discharge. After two days, the number of vesicles increased to 8-10. The woman consulted a local doctor after two weeks and for which she was prescribed anti-allergens and local application. It healed within 2 – 4 days with crusting and hyperpigmentation. She was temporarily relieved of her symptoms, yet similar lesions started developing on her abdomen, back, armpit, neck, medial aspect of the thigh, and groin, and she has taken treatment for the same. The woman reports saying that her father had the same problem, but no history of fever.

Later on, she has observed the increase in the size of the vesicles to bulla and burst forming, and shallow ulcers with subsequent lesions. The episodes of lesion occurrence gradually increased every second month. She consulted a dermatologist in tertiary care hospital and prescribed Tab Bactrim DS (BD) Tab Clotrimazole 800mg (BD), Tab Hifinac (SOS), and Clonate F cream (local application). The lesions were completely healed, and there was a significant improvement in the lesions. Symptoms were reduced temporarily with the treatment but were not cured; instead, she had episodes of similar legions occurring on her body on and off. . The woman tried many oral and topical antibiotics, corticosteroids, which resulted in partial or temporary improvement.

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Manuscript received: November 10, 2019
Revision accepted: February 24, 2020.

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How to cite this article: Ansuya, Shashidhara Y N. (2020). A rare case: Hailey-Hailey disease. *Manipal Journal of Nursing and Health Sciences*, 6(1), 46-49.

There was not much overall improvement in her condition that later started steadily worsening.

After two years, she observed plenty of watery, itchy vesicles spread over the body with pain in the lesion and got admitted to tertiary care hospital. All her blood and urine investigations were normal. Pus from vesicle was sent for culture and sensitivity and obtained positive report for staphylococcus aureus moderate growth. A 3.5 mm lesion biopsy was sent for histopathology. On histopathological examination, a focally ulcerated hyperkeratotic stratified squamous epithelium with subcorneal and suprabasal bullae, neutrophilic exocytosis was reported. Supra basal acantholysis and blunting of rete ridges overlying upper dermis showing perivascular and perianoadnexal lymphocytic inflammatory infiltrate and revealed to be Hailey-Hailey disease. She was treated with Injection Tricot 40 mg stat dose (corticosteroid to reduce swelling/ lesion and pain, itching), Tab DoxyTis LB 100 mg OD (to reduce bacterial infection), Propy dream cream (to minimize the symptoms of inflammation such as redness, swelling, itching), and KZ soap (to reduce fungal infection). In a few days, the lesions started responding to the treatment and started healing with crusting and hyperpigmentation. She was discharged on the 5th day of hospitalization with advice to take the tablets regularly as prescribed and apply the cream daily as mentioned above, and she was also instructed to take a bath regularly with prescribed soap.

During the home visit, she had expressed that she felt better with treatment on being provided with a cold compress, daily savlon dressing, and applied prescribed cream. She was advised to adopt lifestyle modifications i.e., to wear soft and loose cotton dress and underwear and triggering factors such as excessive heat, excessive sweating, and friction by any cause. She had to focus more on skincare, personal hygiene, diet, and weight management to avoid the physical activity that involves friction against the surface of the skin. She followed the advice given and consulted a dermatologist for the follow-up. There was rigorous improvement seen after one month. The lesions were healed, and post-lesional hyperpigmentation was found after this treatment (figure1 & figure 2). The photo was taken after getting consent from the woman. The frequency of lesions decreased, and itchy lesions were most frequent during the summer season.

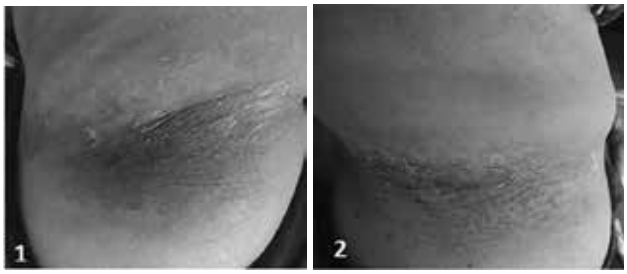

Figure 1 and 2: Photographs showing post-lesional hyperpigmented plaques over the abdomen

Discussion

The first case was described in 1939 by Hailey- Hailey brothers. HHD is also known as familial benign chronic pemphigus and considered a rare disease in India. HHD caused due to genetic mutation in the ATP2C1 gene that generates a protein. This protein is essential to maintain skin health. This condition becomes evident after puberty, usually, appearing to people in the age group of 30 – 40 years, but symptoms can develop at any age. As per reports, HHD affects one in fifty-thousand people. HHD frequently goes misdiagnosed or undiagnosed. This makes the skin disorder hard to detect, and therefore it is hard to determine the actual frequency in the common population (Burgdorf et al., 2010).

The gene ATP2C1 contains directions for the production (encoding) of a protein in the cells that functions as a pump for calcium and magnesium. (Xu, Shi, Diao, Jiang, & Xiao, 2017).

Calcium ions play an essential role in adhesion between cells. The protein is most abundant in keratinocytes, the primary cell type of the outermost layer of skin (epidermis). Failure of keratinocytes to stick together results in the blistering seen in the disease (Calonje, Brenn, Lozar, Alexander, & McKee, 2012).

HHD is a genetic skin disease. The danger of transferring the defective gene from a HHD affected parent to the next generation is 50% for every pregnancy. The risk is identical for males and females (Vasudevan et al., 2015). In terms of symptoms, emotions, and social functioning, and psychological well-being, HHD affects the quality of life (Gisondi, Samponga, Annessi, Girolomoni, & Abeni, 2005).

It is difficult to plan a treatment protocol for HHD. As per the reports, there is no complete cure for HHD. The fundamental genetic flaw cannot be altered. However, the treatment does help patients, and long remissions are common. The treatment goal should be focused to relieve the patient from painful symptoms and improve general health. A concoction of topical antibiotics and antifungal agents along with systemic, topical, and intralesional corticosteroids are useful in the management of HHD in many cases. Oral Acitretin (second-generation retinoid), 25 mg once daily, was prescribed for HHD. In 4 weeks, the lesions started to respond, and after 3 months, there was complete regression. Acitretin proceeded uninterrupted for a total of 5 months, and the patient was monitored once a month, including full hemogram, liver and renal function tests, and serum lipid profile, with clinical review and laboratory examinations. Without any side effects, she tolerated the drug well and was followed up for 2 years without any relapse (Vasudevan et al., 2015).

Ibrahim conducted an observational study among three HHD patients to find the effectiveness of low-dose naltrexone hydrochloride in the treatment of HHD. The study reported that there was a great reduction in the symptoms of Hailey-Hailey disease after the intervention. Due to the improvement in the condition, there was an improvement in the quality of life and also reduced the symptoms of depression (Ibrahim, Hogan, Vij, & Fernandez, 2017).

A study reported that in combination with topical pimecrolimus and topical antibiotics, the Hailey-Hailey patient was treated with Acitretin 0.75 mg/kg per day. However, the patient's condition deteriorated, and the medicine was switched to cyclosporine 2.5 mg/kg per day a month later. Within a week of cyclosporine treatment there was 80% clearance of skin lesions with complete clearance in three weeks. The dose of cyclosporine was tapered off by 0.5 mg/kg and was stopped over six months. Two years later, during a follow-up, the patient was in stable health with only mild recurrences that could be managed with adopting good skin care daily (Nanda et al., 2010).

Botulism type A may be an effective and safe non-surgical alternative for the treatment of HHD in intertriginous areas such as the axillae. It is a very safe and low transient side effects (Lapiere et al., 2000).

Conclusion

Although the course and progress of the disease is unpredictable, its severity can be reduced with treatment, nursing care, and lifestyle modification. Nursing care at home setting will be tremendously beneficial in reducing the problem. A health care professional should have knowledge regarding HHD to identify at the early stage, provide care at the home setting, and refer to the hospital/health center for the treatment. It is essential to educate patients suffering from the disease along with their family members about HHD to avoiding aggravating factors.

Sources of support: None
Conflict of interest: None declared
Source of support in the form of grants: None

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