Hailey-Hailey disease (Familial benign chronic pemphigus, FBCP) was first described in 1939[1]. It is a dominantly inherited disorder with a family history in 70% of the patients. This condition is caused by mutation in the ATP2C1 gene[2], which encodes a calcium pump localized in the Golgi apparatus. The impaired calcium homeostasis results in abnormalities in the adhesion molecules causing acantholysis of keratinocyte.

The condition most commonly begins after puberty, the highest prevalence is in the third to fourth decade. Both sexes are affected equally. Although the primary lesion is a vesicle arising on normal skin, maceration is the usual presentation which occurred mainly on the neck, axillae, inframammary fold, groin, flexural and opposing surface, but may be widespread.[3] Rarely, there is involvement of the cornea, conjunctiva, oral mucosa, esophagus, and genitalia. The active inflammatory border spreads peripherally and heals without scarring. As these macerated areas become colonized and infected, they develop a fetid odor. The lesions may be associated with pruritus, tenderness, burning and pain on motion. This condition is exacerbated by friction, sweating, heat, irritants, wearing tight clothing, stress and premenstruation.

Histologic examination showed suprabasilar cleavage and acantholysis which give the characteristic dilapidated brick wall appearance of epidermis. There is mild superficial perivascular lymphocytic infiltration in upper dermis. A negative direct and indirect immunofluorescence helps to differentiate from pemphigus vulgaris.

Treatment has never been assessed in a large group of patients although topical corticosteroid and topical or oral antibiotics are generally recommended.[4] Systemic corticosteroid, cyclosporin, oral retinoids, dapsone, and methotrexate have been used in severe case. Topical tacrolimus, dermabrasion, photodynamic therapy with 5-aminilevulinic acid, CO_2 laser vaporization, excision and grafting have been shown to be effective in some cases[5].

References:

1. Hailey J, Hailey H. Familial benign chronic pemphigus. Arch Dermatol Syphilol 1939; 39: 679-85.
2. Dobson SC, Fairclough RJ, Dunne E, Brown J, Dissanayake M, Munro CS, et al. Hailey-Hailey disease: Molecular and clinical characterization of novel mutations in the ATP2C1 gene. J Invest Dermatol 2002; 118(2): 338-51.
3. Marsch WC, Stuttgen G. Generalized Hailey-Hailey disease. Br J Dermatol 1978; 99: 553-60.
4. Burge SM. Hailey-Hailey disease: The clinical features, response to treatment and prognosis. Br J Dermatol 1992; 126: 275-82.
5. Fitzpatrick TB, Ortonne JP. Hailey-Hailey disease. In: Freedberg IM, Eisen AZ, Wolff K, Austen KF, Goldsmith LA, Katz SI. Fitzpatrick's dermatology in general medicine vol. 1, 6th eds. New York: Mcgraw-Hill, 2003: 622-4.

CASE 3

A 57-year-old female from Samutprakan

Chief complaint:

She developed localized upper lip swelling for 1 year

Present illness:

One year ago, she developed asymptomatic, localized swelling at her upper lip.

Past history:

She refused history of facial or lip cosmetics and previous lip trauma. She was a known case of diabetes mellitus type 2 for 10 years and on oral hypoglycaemic drugs.

Physical examination:

General appearance A young female patient, cooperative
HEENT. Not pale, not icteric
Oral mucosa – No scrotal tongue
No facial palsy
Upper lip – Illed defined erythematous, indurated, firm to soft plaque at upper lip

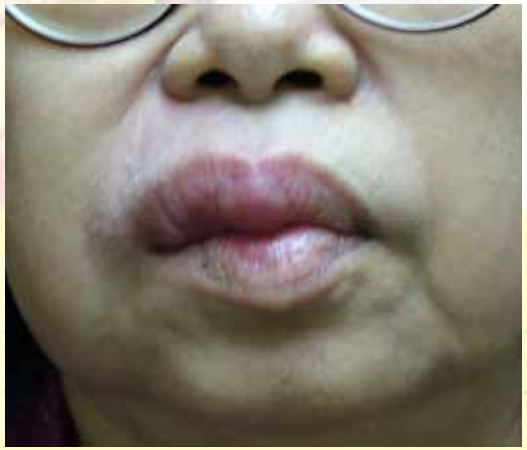

Fig. 3.1

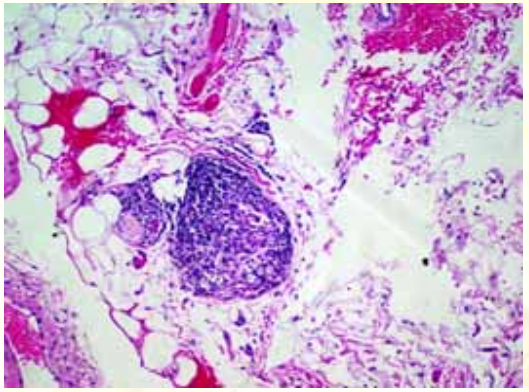

Fig. 3.2

Histopathology: (S05- 17121)

Nodular infiltration of lymphocytes, histiocytes with tuberculoid granuloma with parakeratosis and epidermal hyperplasia

Diagnosis: Cheilitis granulomatosa
Presenter: Sarawut Boonpasat
Consultant: Natta Rajatanavin
Treatment: Kenacort IL 10 mg/mL 0.2 mL

Discussion:

Cheilitis granulomatosa (Meischer's cheilitis) is a chronic swelling of the lip due to granulmatous inflammation of unknown cause. There is no male or female preponderance. It occurs most common at upper lip, lower lip and both lip, respectively. It slowly and progressively swells. Cranial nerve palsy occurs only 20% (usually facial nerve, unilaterally). Rarely other cranial nerves may be affected (olfactory, auditory, glossopharyngeal, hypoglossal and vagus nerve). It may ocassionally be associated with sarcoidosis or Crohn disease and some may develop regional ileitis even years latter. Melkersson-Rosenthal syndrome compose of

1. Cheilitis granulomatosa
2. Scrotal tongue
3. Facial palsy