Kindler Syndrome: A Rare Genodermatosis Presenting in 2 Brothers

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Case Report

We report a case of two Asian brothers, aged 21 and 29, born to consanguineous parents, who presented with cutaneous hyper- and hypopigmentation, skin atrophy, and skin fragility of the face, neck, and distal parts of the extremities. They were the first and second, full-term pregnancies in a family without any history of skin disease. Both patients had a history of recurrent minor trauma-induced blistering over the extensor aspects of the forearms, lower legs, dorsal hands, and feet since infancy. The blisters contained either serous or hemorrhagic fluid and typically ruptured within 4 to 5 days producing erosions that healed with dyspigmentation and atrophy, but no scarring. The tendency for blister formation after minor trauma gradually subsided by the ages of 13 and 15, respectively. Additionally, the brothers had severe skin fragility affecting the face, neck, distal limbs, and gums. They reported increased photosensitivity with sunburn after minimal sun exposure and exacerbation of their disease during the summer months. Dysphagia started at the ages of 17 and 19 and has been persistent with variable severity. The evolution of the disease led to the subsequent development of reticular pigmentation, progressive skin atrophy, gingival fragility with bleeding, webbing of the fingers and toes, nail dystrophy, dysuria and eye soreness.

On physical examination, both patients had normal physical and mental development. Skin examination revealed poikiloderma, patchy hyperpigmentation and hypopigmentation, and telangiectasias (Figures 1 and 2). Also, skin fragility was noted mainly on the face, neck, upper chest, and over the dorsal hands and feet.

Figure 1: Patchy hypo- and hyperpigmentation with poikiloderma over the back
Figure 2: Patchy hyperpigmentation with poikiloderma on the upper chest and neck

There was no evidence of scarring. Most of the nails were rough, discolored and showed longitudinal ridging (Figure 3). Dermatoglyphic patterns were lost on the palms and soles (Figure 4). Examination of the oral cavity showed poor dentition, gingival swelling with easy bleeding, hyperpigmentation of the lips, and synechiae between the lips and the gums (Figure 5).

Figure 3: Yellowish discoloration and longitudinal ridging of the bilateral thumbnails

Figure 4: Loss of dermatoglyphic pattern on the palms

Figure 5: Hyperpigmentation of the mucosal lips with gingival swelling

Ophthalmologic evaluation revealed sparse eyelashes and ectropion of the lower eyelids. Blood tests, urine analysis and stool examinations were normal. An EGD performed on one of the brother’s demonstrated esophageal stenosis, which was subsequently surgically corrected.

Skin biopsies demonstrated epidermal atrophy, flattening of the rete ridges, basal layer focal vacuolization and degener-
Kindler Syndrome

Kindler Syndrome is a rare autosomal-recessive genodermatosis that presents with congenital poikiloderma, characteristic bullae, and several pre-malignant and malignant conditions. Initially described by Theresa Kindler in 1954, it is characterized by cutaneous atrophy, acral blister distribution, and squamous cell carcinoma. The pathophysiology of Kindler syndrome involves abnormal skin fragility disorder involving the extracellular matrix binding to actin, instead of keratin. Kindlin-1 is a signaling protein involved in actin cytoskeleton linking to the extracellular matrix, and its mutation (KIND1) was discovered in a Native American tribe.

In 2003, a loss-of-function mutation was mapped to a gene on 20p12.3 and named KIND1 (encoding kindlin-1). The KIND1 mutation was found in patients with Kindler syndrome, and screening for this mutation can help in the diagnosis. Genetic analysis can rule out dystrophic epidermolysis bullosa.

Management of Kindler syndrome is mainly supportive and preventative. Trauma should be avoided to reduce blister formation, and sun avoidance may help with poikiloderma. Excellent wound care should be practiced to avoid infections of bullae. Since patients with Kindler syndrome are prone to esophageal, anal, and urethral stenosis, surgical procedures may be necessary to correct these abnormalities.

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References