Dermatology Practical & Conceptual

A Rare Genetic Disorder: Keutel Syndrome with Dermatologic Manifestation

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Case Presentation

A 26-year-old female presented with erythematous atrophic irregularly bordered depressed patches on both sides of the neck that gradually developed over the 5 months. She was previously diagnosed with contact dermatitis, and treated with two weeks of topical hydrocortisone cream without any improvement. She also had stridor and recurrent respiratory infections since her childhood, hear-loss, and nose augmentation operation 6 years ago. Dermatological examination revealed erythematous atrophic patches on both sides of the neck, and laxity on both sides of the trunk (Figure 1, A-C). In addition, frontal bossing, maxillary hypoplasia, hypoplastic small nose with a broad, depressed nasal bridge and hypoplastic nostrils, short philtrum, prominent eyes, and dental malocclusion were observed. Her parents were first-degree cousins. On presentation to our clinic, punch biopsy from multiple skin lesions revealed elastin fragmentation and loss on the superficial dermis (Figure 1 D). Based on these findings our patient was diagnosed with Keutel syndrome with skin manifestation. She was treated with barrier cream and sunscreen.

Teaching Point

Keutel Syndrome (KS) is a rare autosomal recessive hereditary genetic disorder with an estimated prevalence of 1:1000000 [1]. It is associated with loss-of-function mutations in the gene coding for the matrix Gla protein (MGP). Abnormal calcification of cartilaginous tissues resulting in malformations of skeletal tissues (eg, midface hypoplasia), respiratory defects (eg, tracheobronchial cartilage calcification), and cardiovascular defects (eg, arterial calcification). Khosroshahi et al reported long-term follow-up results of 4 patients with KS [2]. All patients developed erythematous atrophic skin lesions after 30 years of age. The pathogenesis of skin findings in KS is not fully understood. It is thought to develop because of the destruction of elastic fibers in the papillary dermis due to progressive abnormal calcification [1,2]. However, we think that further studies will show the reasons why especially the elastic fibers of the papillary dermis are affected and that these findings develop after the age of 30. Although systemic treatment with vitamin K supplements has been tried previously, curative treatment has not been available [3]. However, multidisciplinary follow-up for

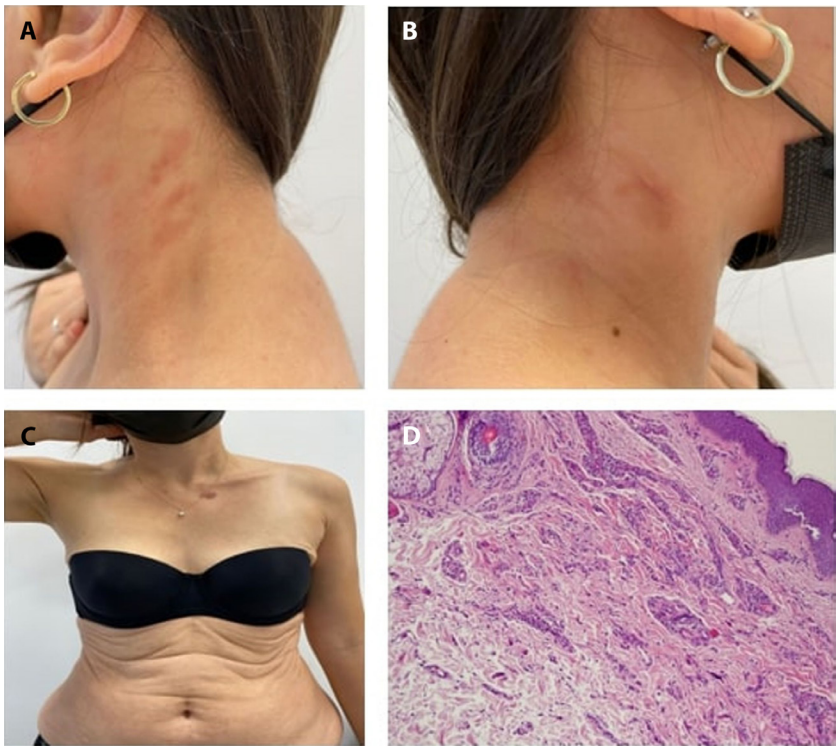

Figure 1. (A, B) Erythematous atrophic patches on the both sides of neck. (C) Prominent laxity on the trunk, (D) Fragmantation and loss of elastin on the dermis (H&E, X100).

systemic involvement is vital [1,2]. Dermatologists play a key role also in the diagnosis of genetic diseases. Keutel syndrome is a rare genetic disorder but should be kept in mind, especially in the presence of a phenotype suggestive of a genetic syndrome and erythematous atrophic patches located on the neck and upper body.

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