Weber-Cockayne type epidermolysis bullosa simplex associated with a novel mutation in keratin 5 and amyloid deposits

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摘要

Abstract

Epidermolysis bullosa simplex (EBS) encompasses a group of hereditary bullous diseases characterized by intraepidermal blister formation caused by mutations of keratin 5 (KRTS) or keratin 14 (KRT14) genes. Weber-Cockayne (EBS-WC: OMIM 131800)is the most common type, which is characterized by localized formation of blisters and is usually confined to the acral/friction area. We report a patient with EBS-WC with a novel mutation in KRTS (Val to Ala) at position 324. Interestingly, globules of amyloid deposition were identified in the papillary dermis; this has never previously been reported in a patient with EBS.