Epidermolysis bullosa hereditaria simplex. Case report.

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SUMMARY

Blistering is a common childhood phenomenon. There is a group of heterogeneous, genetic mechanobullous diseases termed epidermolysis bullosa (EB) whose hallmark is blistering due to minor trauma or pressure. Some patients with EB may be limited in common extracurricular activities such as playing tennis or hiking, while others may be asymptomatic with occasional blistering. The level of vesiculation within the skin defines three major subtypes of EB: EB simplex, dystrophic EB and junctional EB. We describe a patient and review the simple type of epidermolysis bullosa (EBS), which has an incidence of approximately 10-30 cases per million. The incidence of EBS is most probably higher than the stated figure, due to underreporting of mild cases. The severity of EBS depends upon the type of underlying defect.

Introduction

Skin blistering is a relatively frequent manifestation in dermatological disorders of childhood. Blister formation may be observed in infectious diseases such as impetigo or primary varicella, in certain acquired immunological conditions such as linear IgA dermatosis or dermatitis herpetiformis Duhring and in hereditary diseases such as incontinentia pigmenti Bloch-Sulzberger and hereditary bullous epidermolysis (EBs).

EBs are subdivided into three groups according to their pathogenetic mechanisms. *Dystrophic epidermolysis bullosa (DEB)* includes seriously disabling conditions characterized by subepidermal blister formation. Blisters commonly heal with scarring and milia formation. The pathogenesis of DEB has been attributed to varying degrees of faulty Type VII collagen, which results in deficient anchoring fibrils of the basement membrane zone. There are four major forms of DEB that make up the majority of cases. Rare variants, which usually produce severe disease, also exist. The two autosomal dominant DEBs are the Cockayne-Touraine variant and the Pasini variant. The two autosomal recessive forms of DEB are localized recessive DEB (RDEB) and generalized RDEB. RDEB is transmitted recessively and inlcudes a variable spectrum of clinical severity. The Hallopeau-Siemens variant, or HS-RDEB, is notable due to its severely deforming course. The disease begins

KEY WORDS

blistering disorder, epidermolysis bullosa, simplex, hereditary, child