Epidermolysis bullosa simplex due to exophilin 5 deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Epidermolysis bullosa simplex due to exophilin 5 deficiency. ORPHA:412189

Epidermolysis bullosa simplex due to exophilin 5 deficiency is a rare, hereditary, basal epidermolysis bullosa simplex characterized by mild, generalized trauma-induced scale crusts and intermittent blistering, sometimes combined with erosions and bleeding, recovering with slight scarring and post-inflammatory hyperpigmentation. Clinical symptoms improve with age.