

Opis choroby *

Definicja

Exfoliative ichthyosis is an inherited, non-syndromic, congenital ichthyosis disorder characterized by the infancy-onset of palmoplantar peeling of the skin (aggravated by exposure to water and by occlusion) associated with dry, scaly skin over most of the body. Pruritus and hypohidrosis may also be associated. Well-demarcated areas of denuded skin appear in moist and traumatized regions and skin biopsies reveal reduced cell-cell adhesion in the basal and suprabasal layers, prominent intercellular edema, numerous aggregates of keratin filaments in basal keratinocytes, attenuated cornified cell envelopes, and epidermal barrier impairment.

Dane

Klasyfikacja

Choroba

Synonimy

Autosomal recessive exfoliative ichthyosis

Autosomalna recesywna rybia łuska złuszczająca

Ichthyosis exfoliativa

Kod ORPHA

289586

Kod OMIM

617115

Kod ICD10

Q80.8

Kod ICD11

EC20.02

*Źródło

orphanet