Opis choroby *

Definicja

A group of rare inherited non-syndromic ichthyoses characterized by mutations in keratin genes. Mutations in KRT1 and KRT10 cause most cases of epidermolytic ichthyosis (EI), as well as congenital reticular ichthyosiform erythroderma (CRIE). EI manifests at birth with generalized blistering, which later transforms into hyperkeratosis. Severe palmoplantar involvement is suggestive of the presence of a KRT1 mutation. CRIE patients present at birth with erythroderma and scaling, often with a collodion membrane, and gradually develop confetti-like clear areas of normal skin. KRT2 mutations are associated with superficial epidermolytic ichthyosis (SEI), which is clinically similar to EI, but generally milder and more localized.

Dane

Klasyfikacja
Grupa fenomenów

Synonimy
KPI
KPI

Kod ORPHA
281103

Kod OMIM
-

Kod ICD10
-

Kod ICD11
EC20.03

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*Źródło

orphanet