Opis choroby *

Definicja

A rare, inherited, non-syndromic ichthyosis characterized by congenital, generalized erythroderma with cutaneous blistering and erosions, resembling collodion presentation at birth, replaced by progressive hyperkeratosis later in life without palmoplantar involvement. The ultrastructural pathology consists of sparse keratin filaments and keratin clumps that show a nearly homogeneous, amorphous structure.

Dane

Klasyfikacja
Choroba

Synonimy
AREI
AREI

Kod ORPHA
512103

Kod OMIM
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Kod ICD10
Q80.8

Kod ICD11
EC20.03

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*Źródło

orphanet