

Opis choroby *

Definicja

Harlequin ichthyosis (HI) is the most severe variant of autosomal recessive congenital ichthyosis (ARCI; see this term). It is characterized at birth by the presence of large, thick, plate-like scales over the whole body associated with severe ectropion, eclabium, and flattened ears, that later develops into a severe scaling erythroderma.

Dane

Klasyfikacja

Choroba

Synonimy

HI

Rybia łuska płodowa, typu harlekin

Rybia łuska wrodzona, typu harlekin

Ichthyosis congenita, Harlequin type

Autosomal congenital ichthyosis, Harlequin type

Kod ORPHA

457

Kod OMIM

242500

Kod ICD10

Q80.4

Kod ICD11

EC20.02

[*Źródło](#)

orphanet