

## 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