

# Rybia łuska wrodzona - małogłowie - tetraplegia

## Kod Orpha: 2271 Kod OMIM:

### Opis choroby \*

#### Definicja

A rare autosomal ichthyosis syndrome with prominent neurologic signs characterized by the association of congenital ichthyosis with severe developmental delay, microcephaly, spastic tetraplegia, sensorineural hearing impairment, athetosis, and myoclonus. Marked epileptic discharges with occurrence of tonic spasms have also been reported. Cerebral MRI shows diffuse cortical atrophy. There have been no further descriptions in the literature since 1995.

#### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Congenital ichthyosis-microcephalus-quadriplegia syndrome  
Rybia łuska wrodzona - małogłowie - kwadriplegia

#### Kod ORPHA

2271

#### Kod OMIM

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#### Kod ICD10

Q87.8

#### Kod ICD11

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#### [\\*Źródło](#)

orphanet

### Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.