

# Dermatoleukodystrofia

**Kod Orpha: 1659 Kod OMIM: 221790**

## Opis choroby \*

### Definicja

A rare leukodystrophy characterized by congenital thickened, wrinkled skin showing loss of elasticity, in combination with childhood onset of rapidly progressive generalized cognitive and motor impairment quickly resulting in a vegetative state and early death.

Neuropathologic examination reveals neuroaxonal leukodystrophy with numerous neuroaxonal spheroids and diffuse loss of axons and myelin sheaths.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Cutis laxa-leukodystrophy

#### Kod ORPHA

1659

#### Kod OMIM

221790

#### Kod ICD10

E75.2

#### Kod ICD11

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### [\\*Źródło](#)

orphanet

## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.