

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 | Synonimy | |
|------------------|---------------------------|-----------|
| Choroba | Cutis laxa-leukodystrophy | |
| Kod ORPHA | Kod OMIM | Kod ICD10 |
| 1659 | 221790 | E75.2 |
| Kod ICD11 | | |
| - | | |

*[Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.