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
-

*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.