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