

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
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Kod ORPHA 1659	Kod OMIM 221790	Kod ICD10 E75.2
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Kod ICD11

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*Źródło

orphanet