Autosomalna recesywna luźna skóra typu 2A
Kod Orpha: 357058 Kod OMIM: 278250

Opis choroby *

Definicja
A rare, genetic, dermis elastic tissue disease characterized by redundant, overfolded skin of variable severity, ranging from wrinkly skin to cutis laxa associated with pre- and post-natal growth retardation, hypotonia, mild to moderate developmental delay, late closure of anterior fontanelle, and craniofacial dysmorphism (including microcephaly, hypertelorism, downslanting palpebral fissures, large, prominent nasal root with funnel nose, small, low-set ears, long philtrum, drooping facial skin). Additional manifestations may include seizures, intellectual disability, congenital hip dislocation, inguinal hernia, and cortical and cerebellar malformations. Pretibial pseudo-ecchymotic skin lesions have occasionally been associated.
Dane

Klasyfikacja
Choroba

Synonimy
ARCL2A
ARCL2A

Kod ORPHA
357058

Kod OMIM
278250

Kod ICD10
Q82.8

Kod ICD11
LD28.2

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*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.