Opis choroby *

Definicja

A spectrum of connective tissue disorders characterized by the association of wrinkled, redundant and sagging inelastic skin with growth and developmental delay, and skeletal anomalies. The spectrum ranges from patients with classic autosomal recessive cutis laxa type 2 (ARCL2, Debré type) to patients with a milder form of the disease, wrinkled skin syndrome (WSS).

Dane

Klasyfikacja
Grupa fenomenów

Synonimy
ARCL2
ARCL2
Skóra wiotka z luźnością w stawach i opóźnieniem w rozwoju
Cutis laxa with joint laxity and developmental delay

Kod ORPHA
90350

Kod OMIM
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Kod ICD10
Q82.8

Kod ICD11
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*Źródło

orphanet