

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