Opis choroby *

Definicja

A generalized connective tissue disorder characterized by the association of wrinkled, redundant and sagging inelastic skin with severe systemic manifestations (lung atelectesias and emphysema, vascular anomalies, and gastrointestinal and genitourinary tract diverticuli).

Dane

Klasyfikacja
Choroba

Synonimy
ARCL1
Autosomalna recesywna skóra wiotka with severe systemic involvement
ARCL1
Autosomalna recesywna skóra wiotka z rozedmą płuc
Autosomal recessive cutis laxa with severe systemic involvement
Autosomal recessive cutis laxa, pulmonary emphysema type

Kod ORPHA
90349

Kod OMIM
219100

Kod ICD10
Q82.8

Kod ICD11
LD28.2

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*Źródło

orphanet