

Opis choroby *

Definicja

A generalized connective tissue disorder characterized by the association of wrinkled, redundant and sagging inelastic skin with severe systemic manifestations (lung atelectasias 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

*Źródło

orphanet