

Opis choroby *

Definicja

A rare, genetic, dermis elastic tissue disorder characterized by generalized cutis laxa associated with severe, usually early-onset, pulmonary emphysema, frequent and severe gastrointestinal and genitourinary involvement (i.e. bladder/intestine diverticula and/or tortuosity, gastrointestinal fragility, hydronephrosis), and mild cardiovascular involvement (typically limited to peripheral pulmonary artery stenosis only).

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

ARCL1C

Zespół Urbana, Rifkina i Davisa

Autosomal recessive cutis laxa type 1C

Urban-Rifkin-Davis syndrome

Kod ORPHA

221145

Kod OMIM

613177

Kod ICD10

Q82.8

Kod ICD11

LD28.2

*Źródło

orphanet