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

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*Źródło

orphanet