

## 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