

## Opis choroby \*

### Definicja

A rare multiple congenital anomalies syndrome characterized by variable skeletal abnormalities (including craniostenosis, pectus carinatum, short sternum, joint hyperextensibility, and abnormal vertebrae), cutis laxa with excessive skin folds around the cheek, chin and neck, ambiguous genitalia with a micropenis and perineal hypospadias, an umbilical hernia, intellectual disability, premature aged appearance, and cardiac enlargement involving either the ventricles or atria. Facial dysmorphism is variable and can include multiple hair whorls, ptosis, high and broad nasal root, low set ears and small chin. Enamel hypocalcification, abnormal modelling of tubular bones, and reduced cutis laxa may become apparent later on.

Dane

### Klasyfikacja

Zespół wad wrodzonych

#### Kod ORPHA

3134

#### Kod OMIM

312830

#### Kod ICD10

Q82.8

#### Kod ICD11

LD28.2

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### \*Źródło

orphanet