

## Opis choroby \*

### Definicja

A rare overgrowth syndrome with skeletal involvement characterized by pre- or postnatal onset of overgrowth, accelerated bone age in infancy and early childhood, tall stature, bony overgrowth of the skull base, spondylar dysplasia, and undermodeling of the tubular bones. Facial dysmorphism includes mild hypertelorism, depressed nasal bridge, short and broad nose, and full lower lip. Additional reported features are scoliosis, as well as delayed puberty, cryptorchidism, and hypospadias.

### Dane

### Klasyfikacja

Zespół wad wrodzonych

#### Kod ORPHA

498485

#### Kod OMIM

608811

#### Kod ICD10

Q87.3

#### Kod ICD11

LD2C

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### \*Źródło

orphanet