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