Opis choroby *

Definicja

A rare overgrowth syndrome with skeletal involvement characterized by long and slim body habitus and multiple skeletal manifestations, such as scoliosis, macrodactyly of the big toes, arachnodactyly of fingers and toes, camptodactyly and clinodactyly, and progressive valgus deformities of the feet. Epimetaphyseal dysplasia, bowing of the tibiae, and dysmorphic facial features (hypertelorism, high palate, or micrognathia), as well as aortic root dilatation and umbilical hernia have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
498488

Kod OMIM
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Kod ICD10
Q87.3

Kod ICD11
LD2C

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*Źródło

orphanet