

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	Kod OMIM	Kod ICD10
498488	-	Q87.3
Kod ICD11		
LD2C		

*Źródło

orphanet