Opis choroby *

Definicja

Cleidorhizomelic syndrome is a rhizo-mesomelic dysplasia characterized by rhizomelic short stature/dwarfism in combination with lateral clavicular defects. Additional manifestations include brachydactyly with bilateral clinodactyly and hypoplastic middle phalanx of the fifth digit. X-ray demonstrated an apparent Y-shaped or bifid distal clavicle. Cleidorhizomelic syndrome has been reported in one family (mother and son) and is suspected to be transmitted in an autosomal dominant manner. There have been no further descriptions in the literature since 1988.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Rhizomelic shortness with clavicular defect
Niskorosłość rizomeliczna z wadą obojczyka
Zespół Wallisa, Zieffiego i Goldblatta
Wallis-Zieff-Goldblatt syndrome

Kod ORPHA
1453

Kod OMIM
119650

Kod ICD10
Q77.8

Kod ICD11
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*Źródło

orphanet