

## 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