

## Opis choroby \*

### Definicja

Tibial hemimelia-polysyndactyly-triphalangeal thumb syndrome is a rare, genetic dysostosis syndrome, with marked inter- and intra-familial variation, typically characterized by triphalangeal thumbs, hand and/or foot polysyndactyly and/or absent/hypoplastic tibiae (associated with duplication of fibulae in some cases), although isolated triphalangeal thumbs have also been reported. It is often accompanied with remarkable short stature and additional features may include radio-ulnar synostosis and hand oligodactyly, as well as abnormal carpal and metatarsal bones.

### Dane

#### Klasyfikacja

#### Synonimy

Zespół wad wrodzonych  
Absent tibia-polydactyly syndrome  
Zespół hemimelii piszczelowej, polisyndaktylii i  
kciuka trójpaliczkowego  
Werner mesomelic spectrum

#### Kod ORPHA

988

#### Kod OMIM

188740

#### Kod ICD10

Q87.2

#### Kod ICD11

-

---

#### \*Źródło

orphanet