

## 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

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych	Absent tibia-polydactyly syndrome Zespół hemimelii piszczelowej, polisyndaktylii i kciuka trójpaliczkowego Werner mesomelic spectrum

<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
988	188740	Q87.2

### Kod ICD11

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### \*Źródło

orphanet