

Opis choroby *

Definicja

Radial deficiency-tibial hypoplasia syndrome is a rare, genetic dysostosis syndrome with combined reduction defects of upper and lower limbs characterized by bilateral radial aplasia, absent thumbs and bilateral tibial hypo/aplasia. Additional bone anomalies (including partial toe hypo/aplasia, short fibula and clubhand) may be associated. There have been no further descriptions in the literature since 1996.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

1121

Kod OMIM

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Kod ICD10

Q73.8

Kod ICD11

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*Źródło

orphanet