

Opis choroby *

Definicja

Distal arthrogyposis type 5D is a rare subtype of distal arthrogyposis syndrome characterized by arthrogyposis multiplex congenita affecting the hands, feet, ankle, shoulders and/or neck, with camptodactyly of the fingers and limited knee and hip extension, associated with asymmetric ptosis and, less frequently, other ocular manifestations (e.g. ophthalmoplegia, strabismus). Affected individuals frequently have a bulbous nose, furrowed tongue, micro/retrognathia, a short neck, congenital hip dislocation, club feet, scoliosis and short stature.

Dane

Klasyfikacja

Choroba

Synonimy

DA5D

Artrogrypoza dystalna typu 5 bez oftalmoparezy

Artrogrypoza dystalna typu 5 bez oftalmoplegii

DA5D

Distal arthrogyposis type 5 without
ophthalmoparesis

Distal arthrogyposis type 5 without
ophthalmoplegia

Kod ORPHA

329457

Kod OMIM

615065

Kod ICD10

Q68.8

Kod ICD11

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*Źródło

orphanet