Opis choroby *

Definicja

Distal arthrogryposis type 5D is a rare subtype of distal arthrogryposis syndrome characterized by arthrogryposis 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 arthrogryposis type 5 without ophthalmoparesis
Distal arthrogryposis type 5 without ophthalmoplegia

Kod ORPHA
329457

Kod OMIM
615065

Kod ICD10
Q68.8

Kod ICD11
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*Źródło

orphanet