Opis choroby *

Definicja

Gordon syndrome, also known as distal arthrogryposis type 3, is an extremely rare multiple congenital malformation syndrome characterized by congenital contractures of hand and feet with variable degrees of severity of camptodactyly, clubfoot and, less frequently, cleft palate. Intelligence is normal but in some cases, additional abnormalities, such as short stature, kyphoscoliosis, ptosis, micrognathia, and cryptorchidism may also be present. Gordon syndrome, Marden-Walker syndrome and arthrogryposis with oculomotor limitation and electroretinal anomalies clinically and genetically overlap, and could represent variable expressions of the same condition.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Camptodactyly-cleft palate-clubfoot syndrome
Artrogrypoza dystalna typu 3
Artrogrypoza dystalna typu IIA
Kamptodaktylia - rozszczep podniebienia - stopa końsko-szpotawa
Distal arthrogryposis type 3
Distal arthrogryposis type IIA

Kod ORPHA
376

Kod OMIM
114300

Kod ICD10
Q68.8

Kod ICD11
LD26.4Y

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*Źródło

orphanet