

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

*Źródło

orphanet