

Opis choroby *

Definicja

An inherited developmental defect syndrome characterized by multiple congenital contractures of limbs, without primary neurologic and/or muscle disease that affects limb function, and ocular anomalies (ptosis, external ophthalmoplegia and/or strabismus). Intelligence is normal.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Distal arthrogryposis type 5
Arthrogyroza dystalna typu 5
Arthrogyroza dystalna typu IIB
Arthrogyroza dystalna z oftalmoplagią
Distal arthrogryposis type IIB
Distal arthrogryposis with ophthalmoplegia
Oculomelic amyoplasia

Kod ORPHA

1154

Kod OMIM

108145

Kod ICD10

Q68.8

Kod ICD11

LD26.4Y

*Źródło

orphanet