Opis choroby *

Definicja

A rare sporadic arthrogryposis syndrome characterized by multiple congenital contractures presenting in a very specific pattern. It is typically symmetric, involving all four limbs, with internally rotated shoulders, fully extended and fixed elbows, the wrists fixed in flexion, partially flexed fingers, hips fixed in flexion or extension, adducted or abducted, and sometimes dislocated. The knees may be fixed in extension or flexion, and the feet are usually in severe equinovarus position. The jaw and trunk are relatively spared. Normal limb muscle tissue is replaced by fatty, fibrous tissue.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Amyoplasia congenita
Wrodzona amioplazja

Kod ORPHA
488586

Kod OMIM
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Kod ICD10
Q68.8

Kod ICD11
LD26.41

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*Źródło

orphanet