

Opis choroby *

Definicja

A rare disorder marked by short stature with short extremities (final adult height is 120cm +/- 10cm), and joint malformations leading to multiple joint contractures (principally involving the shoulders, elbows, interphalangeal joints and hips).

Dane

Klasyfikacja

Choroba

Synonimy

Diastrophic dwarfism

Dysplazja diastroficzna

Kod ORPHA

628

Kod OMIM

222600

Kod ICD10

Q77.5

Kod ICD11

LD24.03

*Źródło

orphanet