

Karłowatość z krótkimi palcami typu Mseleni

Kod Orpha: 2619 Kod OMIM: 613342

Opis choroby *

Definicja

A rare and crippling chondrodysplasia, reported mainly in the Maputaland region in northern Kwazulu Natal, South Africa, characterized by a bilateral and uniform arthropathy of the joints that primarily and most severely affects the hip but that can also affect many other joints (i.e. knees, ankles, wrists, shoulders, elbows), and that manifests with pain and stiffness that progressively limits joint movement, eventually compromising a patient's ability to walk. Severe short stature and brachydactyly have been reported in a few patients with MJD.

Dane

Klasyfikacja	Synonimy
Choroba	Mseleni joint disease Choroba stawów Mseleni
Kod ORPHA	Kod OMIM
2619	613342
Kod ICD11	Kod ICD10
LD24.3	Q77.7

*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.