

Opis choroby *

Definicja

Cheirospondyloenchondromatosis is an extremely rare type of enchondromatosis of very early onset (from neonatal period to infancy) characterized by symmetrical multiple enchondromas with metacarpal and phalangeal involvement resulting in short hands and feet, platyspondyly, mild to moderate short stature and intellectual disability.

Dane

Klasyfikacja

Choroba

Synonimy

Generalized enchondromatosis with platyspondyly

Generalized enchondromatosis with platyspondyly

Kod ORPHA

99647

Kod OMIM

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Kod ICD10

Q78.8

Kod ICD11

LD24.2Y

*Źródło

orphanet