Opis choroby *

Definicja

A rare primary bone dysplasia due to matrilin-3 varaints and characterized by disproportionate early-onset dwarfism, bowing of the lower limbs, short, wide and stocky long bones with severe epiphyseal and metaphyseal changes, lumbar lordosis, hypoplastic iliac bones, flat ovoid vertebral bodies and normal hands.

Dane

Klasyfikacja
Choroba

Synonimy
SEMD, MATN3-related
SEMD, typ matryliny-3
SEMD, związana z MATN3
SEMD, matrilin-3 type

Kod ORPHA
156728

Kod OMIM
608728

Kod ICD10
Q77.7

Kod ICD11
LD24.3

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*Źródło

orphanet