Opis choroby *

Definicja

A rare lysosomal disease characterized by dysmorphic features and skeletal changes, restricted joint mobility, short stature, and hand deformities (such as claw hands, stiffness of hands, carpal tunnel syndrome, inability to make fists). Most patients have normal intellectual capacity and the clinical progression is less rapid than that of mucolipidosis type II (MLII).

Dane

Klasyfikacja
Choroba

Synonimy
Pseudo-Hurler polydystrophy
Polidystrofia pseudo-Hurler

Kod ORPHA
577

Kod OMIM
252605

Kod ICD10
E77.0

Kod ICD11
5C56.20

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*Źródło

orphanet