

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 mucopolipidosis 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

*Źródło

orphanet