Opis choroby *

Definicja

A rare, genetic lysosomal storage disease characterized by accumulation of glycosaminoglycans in connective tissue which results in progressive multisystem involvement with severity ranging from mild to severe. The most consistent features include musculoskeletal involvement (particularly dysostosis multiplex, joint restriction, thorax abnormalities, and short stature), limited vocabulary, intellectual disability, coarse facies with a short neck, pulmonary involvement (predominantly decreased pulmonary function), corneal clouding, and cardiac valve disease.

Dane

Klasyfikacja
Choroba

Synonimy
Beta-glucuronidase deficiency
Choroba Sly
MPS7
MPSVII
Mukopolisacharydoza typu VII
Niedobór beta-glukuronidazy
MPS7
MPSVII
Mucopolysaccharidosis type VII
Sly disease

Kod ORPHA
584

Kod OMIM
253220

Kod ICD10
E76.2

Kod ICD11
5C56.3Y

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*Źródło

orphanet