

## 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