

## Opis choroby \*

### Definicja

A lysosomal storage disease with multisystemic involvement leading to a massive accumulation of glycosaminoglycans and a wide variety of symptoms including distinctive coarse facial features, short stature, cardio-respiratory involvement and skeletal abnormalities. It manifests as a continuum varying from a severe form with neurodegeneration to an attenuated form without neuronal involvement.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Hunter syndrome

MPS2

MPSII

Mukopolisacharydoza typu II

Niedobór 2-sulfatazy idorunianu

Zespół Huntera

Iduronate 2-sulfatase deficiency

MPS2

MPSII

Mucopolysaccharidosis type II

#### Kod ORPHA

580

#### Kod OMIM

309900

#### Kod ICD10

E76.1

#### Kod ICD11

5C56.31

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#### \*Źródło

orphanet