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