

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

*Źródło

orphanet