Opis choroby *

Definicja

Mucopolysaccharidosis type 1 (MPS 1) is a rare lysosomal storage disease belonging to the group of mucopolysaccharidoses. There are three variants, differing widely in their severity, with Hurler syndrome being the most severe, Scheie syndrome the mildest and Hurler-Scheie syndrome giving an intermediate phenotype.

Dane

Klasyfikacja
Choroba

Synonimy
Alpha-L-iduronidase deficiency
MPS1
MPSI
Mukopolisacharydoza typu I
Niedobór alfa-L-iduronidazy
MPS1
MPSI
Mucopolysaccharidosis type I

Kod ORPHA
579

Kod OMIM
607016

Kod ICD10
E76.0

Kod ICD11
5C56.30

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*Źródło

orphanet