Opis choroby *

Definicja

Hurler syndrome is the most severe form of mucopolysaccharidosis type 1 (MPS1; see this term), a rare lysosomal storage disease, characterized by skeletal abnormalities, cognitive impairment, heart disease, respiratory problems, enlarged liver and spleen, characteristic facies and reduced life expectancy.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Hurler disease
Choroba Hurler
MPS1H
MPSIH
Mukopolisacharydoza typu 1H
Mukopolisacharydoza typu IH
MPS1H
MPSIH
Mucopolysaccharidosis type 1H
Mucopolysaccharidosis type IH

Kod ORPHA
93473

Kod OMIM
607014

Kod ICD10
E76.0

Kod ICD11
5C56.30

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*Źródło

orphanet