

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	Synonimy
Podtyp kliniczny	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		

*Źródło

orphanet