

Opis choroby *

Definicja

Hurler-Scheie syndrome is the intermediate form of mucopolysaccharidosis type 1 (MPS1; see this term) between the two extremes Hurler syndrome and Scheie syndrome (see these terms); it is a rare lysosomal storage disease, characterized by skeletal deformities and a delay in motor development.

Dane

Klasyfikacja	Synonimy
Podtyp kliniczny	MPS1H/S MPS1H/S MPSIH/S Mukopolisacharydoza typu 1H/S Mukopolisacharydoza typu IH/S MPSIH/S Mucopolysaccharidosis type 1H/S Mucopolysaccharidosis type IH/S

Kod ORPHA 93476	Kod OMIM 607015	Kod ICD10 E76.0
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Kod ICD11
5C56.30

*Źródło

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