

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	Kod OMIM	Kod ICD10
93476	607015	E76.0

Kod ICD11
5C56.30

*Źródło

orphanet