DISEASE:
Hurler-Scheie syndrome

NAME:	Hurler-Scheie syndrome
DESCRIPTION:	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.
ORPHACODE:	93476
SYNONYMS:	MPS1H/S MPSIH/S Mucopolysaccharidosis type 1H/S Mucopolysaccharidosis type IH/S
XREF(S):	Orphanet MedDRA ICD-10 OMIM
ANALYTE(S):	IDUA
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RELATED CONTENT

Related Genetic Tests

- Enzymatic dosage MPS1/Hurler syndrome
- Lysosomal Storage Disease (gene panel)
- Mucopolysaccharidosis (MPS) type I / Hurler-Scheie syndrome

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- alpha-L-iduronidase

Related Gene Panels

- Lysomal Storage (64 genes) - VUB