

DISEASE:
Hurler syndrome

NAME:	Hurler syndrome
DESCRIPTION:	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.
ORPHACODE:	93473
SYNONYMS:	Hurler disease MPS1H MPSIH Mucopolysaccharidosis type 1H Mucopolysaccharidosis type IH
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	IDUA
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: <http://gentest.healthdata.be/disease/3471>

RELATED CONTENT

Related Genetic Tests

- [Enzymatic dosage MPS1/Hurler syndrome](#)

Related Laboratories

- [Centrum Medische Genetica - UZ Brussel VUB](#)

Related Analytes

- [alpha-L-iduronidase](#)

Source URL: <http://gentest.healthdata.be/disease/3471>