

Hurler syndrome

Hurler syndrome, also known as mucopolysaccharidosis type I (MPS I), Hurler's disease, also gargoylism, is a genetic disorder that results in the buildup of glycosaminoglycans (formerly known as mucopolysaccharides) due to a deficiency of alpha-L iduronidase, an enzyme responsible for the degradation of mucopolysaccharides in lysosomes.

Without this enzyme, a buildup of heparan sulfate and dermatan sulfate occurs in the body. Symptoms appear during childhood and early death can occur due to organ damage.

MPS I is divided into three subtypes based on severity of symptoms. All three types result from an absence of, or insufficient levels of, the enzyme α -L-iduronidase. MPS I H or Hurler syndrome is the most severe of the MPS I subtypes. The other two types are MPS I S or Scheie syndrome and MPS I H-S or Hurler-Scheie syndrome.

Hurler syndrome is often classified as a [lysosomal storage disease](#), and is clinically related to Hunter Syndrome. Hunter syndrome is X-linked while Hurler syndrome is autosomal recessive.

It is named for Gertrud Hurler (1889–1965), a German pediatrician.

see [myelopathy](#).

Progressive neurocognitive decline in this condition can be accompanied by [macrocephaly](#), [ventriculomegaly](#), and/or periventricular signal changes on MRI, which often leads to a neurosurgical referral.

In a case, Liang and Singhal describe a 2-year-old boy with ventriculomegaly and periventricular T2 signal changes, both of which decreased following medical management of Hurler syndrome. The authors discuss the possible mechanisms for this finding and the implications for neurosurgical treatment of this condition ¹⁾.

Ziyadeh et al present the first reported case associating MPS-I (Hurler-Scheie subtype) with craniosynostosis. A 2.5-year-old girl presented initially with macrocrania. On clinical and radiological examinations we noted a scaphocephaly with dysmorphic facial features of MPS confirmed later on. Intracranial hypertension was documented at fundoscopy (papilloedema) and ICP monitoring, and then surgically treated. This association of scaphocephaly and MPS-I highlights the importance of a meticulous physical examination performed by craniofacial, metabolic and ophthalmologic teams ²⁾.

Acute hydrocephalus Hurler's syndrome ³⁾.

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Liang J, Singhal A. Regression of ventriculomegaly following medical management of a patient with Hurler syndrome. J Neurosurg Pediatr. 2016 May;17(5):537-9. doi: 10.3171/2015.9.PEDS15477. Epub 2016 Jan 8. PubMed PMID: 26745646.

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Ziyadeh J, Le Merrer M, Robert M, Arnaud E, Valayannopoulos V, Di Rocco F. Mucopolysaccharidosis type I and craniosynostosis. *Acta Neurochir (Wien)*. 2013 Oct;155(10):1973-6. doi: 10.1007/s00701-013-1831-9. Epub 2013 Aug 6. PubMed PMID: 23917744.

Shinnar S, Singer HS, Valle D. Acute hydrocephalus Hurler's syndrome. *Am J Dis Child*. 1982 Jun;136(6):556-7. PubMed PMID: 6807082.

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