

Understanding Mucopolysaccharidosis type I (MPS I)



MPS I disease is a rare, inherited lysosomal storage disorder that is progressive, multisystemic, and can be life-threatening

MPS I is caused by the lack of the lysosomal enzyme α -L-iduronidase. Deficiency of this enzyme results in the progressive accumulation of non-degraded material (called glycosaminoglycans, or GAGs) in cells throughout the body. The accumulated GAGs can cause cardiovascular complications and respiratory disease, which are the leading morbidities associated with early death in patients with MPS I.^{1,2}

MPS I is autosomal recessive and affects males and females equally. The estimated worldwide incidence of MPS I is 1:100,000.¹

Hurler, Hurler-Scheie, and Scheie syndromes

MPS I includes a highly heterogeneous spectrum of severity, signs, symptoms and affected organ systems, with symptom onset often in early childhood.³

Severe MPS I
Rapid progression

Attenuated MPS I
Slower progression



	Hurler	Hurler-Scheie	Scheie
Phenotype			
Median Age of Onset ⁴	6 months	1.8 years	5.3 years
Early and Prevalent Signs ⁴	Hernia, coarse facial features, kyphosis/gibbus	Hernia, corneal clouding, hepatomegaly	Hernia, joint contractures, corneal clouding
Median Age at Diagnosis ⁴	1 year	4 years	9.4 years
Impact on Cognition	Significant mental delay with loss of acquired skills	Normal or slightly delayed, learning disabilities	None

Patient photo courtesy of Dr Guelbert.

NICK, LIVING WITH MPS I (SCHEIE)
Age at first symptom onset: 2 years old
Age at diagnosis: 4 years old

How to identify Signs and Symptoms of MPS I¹⁻⁴



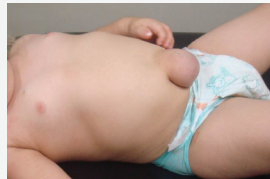
Diagnosis is often delayed since signs and symptoms may be shared with other disorders

Diagnosis may be based on a unique combination of symptoms rather than on a single presenting symptom. Symptom recognition and accurate early diagnosis is important to help facilitate disease management.

Some common differential diagnoses include:

- Juvenile rheumatoid arthritis
- Rheumatoid arthritis
- Idiopathic rheumatoid arthritis
- Other mucopolysaccharidoses
- Multiple sulfatase deficiency
- Arthrogyrosis

Most common signs and symptoms



Recurrent hernia



Poor vision



Poor hand function

If you suspect MPS I disease, refer your patient to a Metabolic Specialist or Geneticist immediately.

For more information about signs and symptoms, visit www.hcp.mps1disease.com



References

1. Neufeld EF, Muenzer J. The mucopolysaccharidoses. In: Valle DL, Antonarakis S, Ballabio A, Beaudet AL, Mitchell GA, eds. *The Online Metabolic and Molecular Bases of Inherited Disease*. New York, NY: McGraw-Hill Education; 2014. Accessed March 03, 2016.
2. Vijay S, Wraith JE. Clinical presentation and follow-up of patients with the attenuated phenotype of mucopolysaccharidosis type I. *Acta Paediatr*. 2005;94(7):872-877. doi:10.1111/j.1651-2227.2005.tb02004.x.
3. Cobos PN, Steglich C, Santer R, Lukacs Z, Gal A. Dried blood spots allow targeted screening to diagnose mucopolysaccharidosis and mucopolipidosis. *JIMD Rep*. 2015;15:123-132. doi:10.1007/8904_2014_308.
4. Cimaz R, Vijay S, Haase C, et al. Attenuated type I mucopolysaccharidosis in the differential diagnosis of juvenile idiopathic arthritis: a series of 13 patients with Scheie syndrome. *Clin Exp Rheumatol*. 2006;24(2):196-202.

Signs and Symptoms



Symptom Presentation	Phenotype	
	Hurler	Hurler-Scheie / Scheie
	Severe Patients	Attenuated Patients
Corneal Clouding	●●●	●●
Stiffened Joints	●●●	●●
Skeletal Abnormalities	●●●	●●
Carpal Tunnel Syndrome	●●●	●●
Cardiac (Valvular) Disease	●●●	●●
Coarse Facial Features	●●●	●
Recurrent Ear, Nose, and Throat Infections	●●●	●
Obstructive Airway Disease/Sleep Apnea	●●●	●
Inguinal or Umbilical Hernia	●●●	●
Growth Deficiencies	●●●	●
Hepatomegaly/Splenomegaly	●●●	●
Spinal Cord Compression	●●●	●
Hearing Loss	●●●	●
Cognitive Impairment	●●●	●
Communicating Hydrocephalus	●●●	—
Abnormally Shaped Teeth	●●●	—

Scale of severity: ●●● most severe to — not present
 For more information, call Sanofi Medical Information at 800-745-4447 (option 2). Photos used with patient permission.

