MUCOPOLYSACCHARIDOSIS TYPE 1 (MPS1)

WHAT IS MPS1?

MPS1 is classified as a lysosomal storage disorder (LSD). Lysosomes are cell organelles that digest and recycle various molecules. Disorders that result in the accumulation of molecules within the lysosomes, including MPS1, are identified as LSDs.

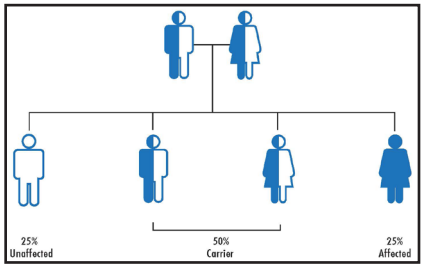
MPS1 is divided into two types: attenuated and severe MPS1 according to the severity of the condition.

THE CAUSE

MPS1 is caused by mutations in the IDUA gene. The normal function of this gene is to produce an enzyme, alpha-L-iduronidase, that breaks down large sugar molecules called glycosaminoglycans (GAGs). A gene mutation results in reduced quantity or complete absence of alpha -L-iduronidase, which causes the accumulation of GAGs within the lysosomes.

INHERITANCE PATTERN

MPS1 is an inherited autosomal recessive (AR) condition. In order to be affected, the individual would have had to inherit one mutation in the gene from each parent. Each parent is referred to as a carrier and does not show symptoms of the condition.



When two carrier parents of an AR condition have children, there is a 25% chance of having an affected child with each pregnancy.

Attenuated MPS1 occurs in approximately 1 in 500,000 newborns, while severe MPS1 occurs in around 1 in 100,000 newborns.

PATIENT CARE

DIAGNOSIS

1. Urine test to look for elevated levels of GAGs.
2. Enzymatic testing to measure alpha-L-iduronidase activity levels in the blood or skin cells.
3. DNA mutation analysis of the IDUA gene.

MANAGEMENT

The management team should include genetics, cardiology, pulmonology, neurology, ENT specialists, audiology, ophthalmology, orthopedics, and developmental specialists.

TREATMENT

The standard of care for patients with severe MPS1 is hematopoietic stem cell transplant (HSCT) which should be done within the first two years of life.

Enzyme replacement therapy (ERT) using laronidase (Aldurazyme) for the missing IDUA enzyme is also available.

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WHAT ARE THE SYMPTOMS?

For patients with severe MPS1, symptoms appear within the first year of life. Those with the less severe or attenuated MPS1 exhibit milder symptoms that appear later in childhood. Listed below are symptoms that may be seen in a patient with MPS1, however symptoms vary from individual to individual, and people with the same disorder may not exhibit all symptoms.

- Macrocephaly
- Enlarged vocal cords causing a hoarse voice
- Hydrocephalus
- Frequent upper respiratory infections/ ear infections
- Heart valve abnormalities*
- Airway obstruction/ sleep apnea*
- Coarse facial features
- Short stature
- Hepatosplenomegaly
- Joint contractures/ skeletal abnormalities

* Most common causes of death

RESOURCES

National MPS Society
mpssociety.org

Gene Reviews: MPS1
ncbi.nlm.nih.gov/books/NBK1162

MPS1 Disease
MPS1Disease.com

Genetics Home Reference
ghr.nlm.nih.gov/condition

Genetic and Rare Diseases
rarediseases.info.nih.gov

Supported through an educational grant (IME-2018-13220) from Sanofi Genzyme