

Frequently Asked Questions

- 1. If no members of my family are affected by MPS, can I still have a child with the condition?**

Yes. Due to the way the disease is inherited, it is possible for the defective gene to be present in the family without anyone showing symptoms (carriers). Hence, you may not be aware of a family history.
- 2. If I am a carrier, can I develop MPS?**

No. Carriers are healthy people who do not manifest any symptoms.
- 3. Since MPS is a genetic disorder, is it possible to have more than one child with the condition?**

Yes. If both parents are carriers, there is a 25% chance of having an affected child with each pregnancy for all types of MPS except MPS II. For MPS II, there is a 50% chance of having an affected son with each pregnancy.
- 4. Would a child with MPS be able to go to school?**

Children with milder symptoms are able to go to school and perform well. However, children with severe MPS will not be able to go to school due to the severity of the symptoms and in some cases mental impairment.
- 5. Can the changes in appearance due to MPS be reversed once treatment is administered?**

No. The changes in facial features are irreversible. Treatment will help stop or delay further changes from happening.
- 6. Can people with MPS achieve a normal lifespan?**

Longevity depends on the type of MPS and its severity. Lifespan ranges from early childhood to middle age. Patients are advised to consult their physician to obtain specific information regarding this.

References

Genzyme MPS Education Handbook

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Rare Disorders Series

Mucopolysaccharidoses
(MPS)

What is Mucopolysaccharidoses (MPS)?

MPS is a group of inherited diseases characterised by the accumulation of harmful amounts of complex molecules in the body.

It is caused by the body's inability to produce certain enzymes that are needed to break down the complex molecules.

The build up of these substances causes progressive damage that affects various organs and impairs physical and mental development.

The absence or deficiency of the required enzymes is due to changes (mutation) in the genes which instruct the production of the specific enzymes.

There are seven types of MPS disorders caused by deficiencies of eleven different enzymes. The features of the different types of MPS vary from one another.

Type	Name of Syndrome
I	Hurler, Hurler-Scheie and Scheie
II	Hunter
III	Sanfilippo
IV	Morquio
VI	Maroteaux-Lamy
VII	Sly
IX	Hyaluronidase Deficiency

All MPS disorders except MPS type II have an autosomal recessive inheritance pattern. This means that to be affected, one needs to have two copies of the chromosome with a gene change.

If both parents are carriers (people having one copy of the mutation) there is a 25% risk of having an affected child with each pregnancy.

MPS type II is inherited via the X-linked inheritance pattern. Males are more likely to develop symptoms than are females.

The sons of females who are carriers each have a 50% chance of having the condition.

The daughters each have a 50% chance of being carriers.

The prognosis for an individual with MPS varies greatly according to the severity of symptoms.

Patients are advised to consult a geneticist to obtain further information on disease progression.

Signs and symptoms

- Coarse facial features
- Clouding of the cornea
- Enlarged head
- Vision and hearing problems
- Chest and back deformities (scoliosis and kyphosis)
- Short and broad hands
- Excess hair growth
- Restricted limb movements
- Short stature

Other ways MPS may affect the body

- Umbilical and inguinal hernia
- Respiratory problems
- Problems with the heart valve
- Mental impairment

Note that symptoms and severity vary according to types of MPS and not all of the symptoms stated above will be seen in an individual with MPS.

Symptoms are usually not present at birth and would manifest during infancy or much later.

Testing

- Clinical examination
Diagnosis of MPS starts off with clinical suspicion. As there are many types of MPS, with overlapping symptoms; collaboration between various specialists is necessary for differentiation.
- Urine test
Performed to look for highly elevated levels of urinary glycosaminoglycans (GAGs)
- Enzyme assay
To investigate enzyme activity in the various tissues in the body. Knowledge of the type of deficient or absent enzyme will allow a diagnosis to be made

Genetic counselling is provided before any genetic testing is done.

Treatment and management

Treatment is given to slow the progression of the disease and to improve quality of life to some extent.

The progressive nature of the disease requires that patients undergo medical evaluation on a regular basis. The systems that need attention are neurologic, ophthalmologic, auditory, cardiac, respiratory, gastrointestinal and musculoskeletal.

Exercising may delay joint problems and improve ability to move.

Enzyme Replacement Therapy (ERT) may aid in improving organ function and mobility. In some centres, bone marrow stem cell transplantation is done for some MPS at an early age (usually before 2 years old)