



MUCOPOLYSACCHARIDOSIS TYPE I (MPS I)

FAMILY FACT SHEET

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 36 hours after birth. The blood is tested for rare disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called Mucopolysaccharidosis type I, also called Hurler syndrome.

A positive newborn screen does not mean your baby has MPS I. It means your baby needs more testing to know for sure.

Your baby's primary care provider or the newborn screening program will notify you to arrange for more testing.

What is MPS I?

Mucopolysaccharidosis type I affects an enzyme in the body that breaks down mucopolysaccharides.

Mucopolysaccharides are large sugar molecules. They are broken down in structures called lysosomes. People who have MPS I do not make enough enzyme needed to break down mucopolysaccharides. Since they are not being broken down, they build up in lysosomes. This build up damages the bones, joints and other tissues.

MPS I is passed on (inherited) from parents to a child. Both the mother and father of an affected child carry a gene change that can cause MPS I. Parents usually do not know they carry the gene change because it does not cause health problems for them.

What problems can MPS I cause?

Mucopolysaccharidosis type I is variable. Some people show symptoms as a baby, called severe MPS I. They will need treatment right away. Others may not show symptoms until later in childhood or adulthood. That is called attenuated MPS I.

A child with MPS I may develop:

- Buildup of fluid around the brain
- Heart valve problems
- Characteristic facial features
- Large liver and spleen
- Clouding of the eye

It is very important to follow the doctor's instructions for testing and needed treatment.

What is the treatment for MPS I?

Mucopolysaccharidosis type I has no cure. Treatment is life-long. Treatment can include:

- Intravenous Enzyme Replacement Therapy (called ERT)
- Bone marrow transplant
- Physical, occupational and speech therapy
- Breathing and dietary treatment

Children who have MPS I should see their regular doctor. They should also see a doctor who specializes in MPS I as part of a special care team.

Prompt and careful treatment helps children with MPS I live the healthiest lives possible.

It is very important to get follow-up testing as soon as possible.

Michigan Resources and Support

Michigan Newborn Screening

Nurse Consultant

Toll-free: 1-866-673-9939

newbornscreening@michigan.gov

Michigan Clinica

Children's Hospital of Michigan 313-832-9330

University of Michigan: 1-734-764-0579

Children's Special Health Care Services

Toll-free: 1-800-359-3722

Online Resources

Medline Plus

<https://medlineplus.gov/genetics/condition/mucopolysaccharidosis-type-i/>

National Organization for Rare Diseases

www.rarediseases.org/rare-diseases/mucopolysaccharidoses