Parent Information
Positive Newborn Screening Test Result for
MUCOPOLYSACCHARIDOSIS TYPE I
(MPS I)

(mew-co-paw-lee-sack-uh-ride-o-sis)

What is Newborn Screening?

After babies are born, five drops of blood are collected from the baby's heel to test for certain diseases. These conditions usually cannot be found simply by looking at or examining a baby.

What is Mucopolysaccharidosis Type I (MPS I)?

MPS I affects an enzyme in the body that breaks down mucopolysaccharides. Mucopolysaccharies are large sugar molecules. They are broken down in structures called lysosomes**.** MPS I is a lysosomal storage disorder. People who have MPS I do not make enough enzyme needed to break down mucopolysaccharides, which build up in lysosomes. This build up damages the bones, joints, heart 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 that can cause MPS I. Parents usually do not know they carry the gene because it does not cause health problems for them. If both parents pass a MPS1 gene to their baby, the baby will develop MPS1 symptoms.

What does a positive result mean?

A positive test means that your baby needs to have more testing to check for MPS I. A positive test does not mean that your baby has MPS I. There are other reasons why a baby might have a positive result. The "screening test" identifies the babies that need more testing. Check with your baby's doctor after additional testing is completed.

What do I need to do now?

Your baby's doctor will tell you how to have the necessary tests done. Testing should be done without any delays even though your baby appears well. If your baby does have MPS I, finding out quickly will allow treatment to be started promptly.

It is also important to follow all of your doctor's instructions for testing and for treatment.

What problems can MPS I cause?

MPS 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: build-up of fluid around the brain, heart valve problems, certain facial features, large liver and spleen, clouding of the eye, and developmental delays.

How is MPS I treated?

Treatment of MPS I includes evaluation and management by a team of specialists who are knowledgeable about the disease. Treatment can include replacing the missing enzyme to help relieve some of the symptoms, but this is not a cure. Stem cell transplant is another possible treatment for patients with MPS I. MPS I has no cure. Treatment is life-long.

Resources for parents

www.babysfirsttest.org
www.dhhs.ne.gov/nsp/

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