When Baby has an abnormal test for Mucopolysaccharidosis type 1 (MPS I)

A small sample of your baby's blood was collected soon after birth and sent to the SC DHEC Public Health Laboratory for testing. This testing is called Newborn Screening. In SC, newborns are screened for several metabolic and genetic disorders.

Sometimes, a second test is needed to help your doctor decide if your baby has one of these disorders. If your baby does have a newborn screening disorder, early treatment will give him or her the best chance to grow up as healthy as possible.

Because an enzyme called Alpha-L-iduronidase (IDUA) was very low or absent in your baby's first test, secondary testing was performed:

Secondary testing indicated your baby may have MPS I.

What is Mucopolysaccharidosis type 1 (MPS I)?

MPS I is an inherited condition that affects many different parts of the body. It is considered a lysosomal storage disorder because people with MPS I have lysosomes (the recycling center of each cell) that cannot break down certain types of complex sugars.

This defect causes undigested sugar molecules and other harmful substances to build up in cells throughout the body, resulting in a variety of symptoms.

Early signs of MPS I include:

- Soft outpouching around the bellybutton (umbilical hernia) or lower abdomen (inguinal hernia)
- Large head (macrocephaly)
- Distinctive facial features that appear "coarse"
- Varying degrees of developmental delay and learning disabilities
- Swollen abdomen (due to enlarged liver and spleen)
- Clouding of the eye (corneal clouding)
- Hearing loss
- Frequent "runny nose"

If your baby shows any of these signs, be sure to contact your baby's health care provider immediately.

How will I know if my baby really has MPS I?

Your baby's newborn screening result showed decreased or absent Alpha-L-iduronidase (IDUA) enzyme activity. You may be referred to a doctor who specializes in this kind of metabolic disorder, called a metabolic specialist or clinical geneticist.

What do I need to do?

Follow your doctor's instructions very carefully.

How is MPS I treated? There are many treatments your baby's health care provider may recommend to minimize the signs of MPS I and help your baby live a healthier life:

Medications

Enzyme replacement therapy (**ERT**) can be an effective treatment for symptoms of attenuated MPS I that do not involve the central nervous system (the brain and spinal cord). This treatment aims to supplement the enzymes that are present at low levels in your baby's lysosomes. ERT may improve growth, joint movement, sleep apnea, respiratory function, pain levels, vision, and liver/spleen enlargement.

If your baby has periods of constipation, your baby's health care provider may recommend mild laxatives to be used very conservatively to relieve discomfort.

Hematopoietic Stem Cell Transplantation (**HSCT**)

Your baby's doctor may recommend your baby receive hematopoietic stem cell transplantation (HSCT) to improve the signs and symptoms of severe MPS I. Hematopoietic stem cells can be found in bone marrow (the spongy tissue inside bones), the bloodstream, or the umbilical cord blood of newborn babies.

Transplanted hematopoietic stem cells are administered through an intravenous (IV) line, much like a blood transfusion. This therapy may reduce facial coarseness, improve hearing and liver/spleen enlargement, stabilize heart function, and slow the decline of cognitive function.

Physical Therapy
Physical therapy is a very important part of treating the signs and symptoms of MPS I. Consistent physical therapy early on can help preserve mobility and lessen pain and joint stiffness.

Surgeries
Your child's health care provider may recommend surgeries to improve your child's quality of life. Removal of the tonsils and adenoids and insertion of ear ventilation tubes (vent tubes) can prevent some upper respiratory infections that may reduce hearing loss.

Hearing aids may be recommended for some individuals. Children with mild to severe MPS I may develop a buildup of fluid in the brain (hydrocephaly). A surgery to relieve the pressure inside the skull may be recommended.

Diet
A dietitian can help you create a nutrition plan to help your baby control diarrhea and constipation, which may occur in those with severe MPS I. There is no diet that can prevent the storage of GAGs because they are actually created by the body.

What else should I do to keep my baby as healthy as possible?

Treating illnesses before they become serious and following your baby's treatment plan carefully are the best things you can do to help your baby grow and develop.

Where can I find more information?

Internet Reference: http://www.babysfirsttest.org/