NORTH CAROLINA NEWBORN SCREENING PROGRAM

Mucopolysaccharidosis I Fact Sheet for Parents

All babies born in North Carolina are screened at birth to look for certain diseases or other health problems that can be treated if caught early. The newborn screening result showed that your baby might have Mucopolysaccharidosis I (MPS I). Your baby will be referred to a specialist for more testing to know for sure.

There are usually no signs of MPS I at birth.

What is MPS I?

MPS I is a rare inherited disorder that affects most parts of the body. Individuals with MPS I cannot break down a group of complex sugars called glycosaminoglycans (GAGs) because they do not have an enzyme called iduronidase. As a result, GAGs build up in cells and cause health problems. Starting treatment for MPS I early has been shown to help prevent, stop, or delay many of the health-related problems.

What are the symptoms MPS I?

Individuals with MPS I have a range of symptoms, or signs of the disease. In severe cases, also known as Hurler syndrome, babies with MPS I can show signs in the first year of life. These symptoms can get worse quickly. In attenuated, or milder cases of MPS I, symptoms may not appear until later in childhood. Symptoms can include:

- Developmental delays (such as delayed speech and walking)
- Large head (macrocephaly)
- Clouding of the eye (corneal clouding)
- Hearing loss
- Frequent runny nose
- Large belly (caused by a large liver and spleen)
- Bony lump on back (spinal kyphosis)
- A large lump or bulge (a hernia) around the belly-button (umbilical hernia) or diaper area (inguinal hernia)

What happens next?

Your baby's pediatrician will refer your baby to a specialist at UNC for follow-up testing. These tests will check your baby's blood and urine to confirm if your baby has MPS I. Three tests may be done. One test will check for the buildup (or high levels) of GAGs (complex sugars) in your baby's urine. A second test will check for a low iduronidase enzyme level. A third test, using blood, will look at your baby's genes (DNA) to check for a gene change (mutation) that causes MPS I. All three of these tests will be used together to confirm if your baby has MPS I.

How is MPS I treated?

Treatment options include:

- **Hematopoietic Stem Cell Transplantation:** Bone marrow or cord blood cells from a donor are given into your baby's vein (IV) so their body can make the missing enzyme. This treatment is recommended for babies with severe MPS I.
- **Enzyme Replacement Therapy (ERT):** ERT is a medication given weekly into your baby's vein (IV) to replace the missing enzyme. This is recommended for babies with attenuated MPS I.

If your baby is diagnosed with MPS I, your baby will be referred to other specialists to help with their care.

Where do I go for more information?

Use your phone's camera to scan the QR codes below.

Baby's First Test

National MPS Society

UNC Pediatric Genetics & Metabolism

NC DEPARTMENT OF HEALTH AND HUMAN SERVICES

State of North Carolina Department of Health and Human Services Division of Public Health

www.ncdhhs.gov

http://slph.ncpublichealth.com

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