NORTH CAROLINA NEWBORN SCREENING PROGRAM

Mucopolysaccharidosis I Fact Sheet for Parents

All babies born in North Carolina are screened at birth 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.

Although there are usually no signs of MPS I at birth, detecting and treating it early during infancy can help prevent or delay many health-related problems.

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 alpha-L-iduronidase (IDUA). As a result, GAGs build up in cells and cause health problems.

What does an abnormal newborn screening result mean?

Newborn screening for MPS I works by doing two tests on your baby's blood sample that was collected from a heel stick after birth, typically at 24 hours of age. First, the newborn screening lab measures the level of your baby's enzyme. If that level is too low, they do a second test to measure your baby's GAG level (also called MPS I marker).

Your baby's newborn screening came back suggestive for MPS I because BOTH of those tests were abnormal.

What are the symptoms MPS I?

Individuals with MPS I can have a range of symptoms depending on how severe their disease is. 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 milder cases of MPS I, known as the attenuated form, symptoms may not appear until later in childhood. Possible symptoms vary and can include the following:

- 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 will be seen by a specialist at the Muenzer MPS Center with UNC Heath Care in Chapel Hill for further evaluation and follow-up testing to confirm if your baby has MPS I. 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 IDUA enzyme level in blood. Additional tests may be necessary depending on your child's newborn screening results and will be coordinated with your providers at the Muenzer MPS Center. For questions, please call the MPS Center (919-228-2432).

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.

Other treatment options may be available and will be discussed by your baby's provider. If your baby is diagnosed with MPS I, your baby will be referred to other specialists to help with their care. Treatment is available here in North Carolina, and the specialists at the UNC Muenzer MPS Center will talk with you about all the treatment options during your visit.

Where do I go for more information?

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

Muenzer MPS Research and Treatment

UNC Pediatric Genetics and Metabolism

Baby's First Test

National MPS Society

Newborn Screening Information Center

Kennedy Ladd Foundation

NC DEPARTMENT OF HEALTH AND HUMAN SERVICES

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

www.ncdhhs.gov

https://slph.dph.ncdhhs.gov/newborn/default.asp

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