NORTH CAROLINA NEWBORN SCREENING PROGRAM

Mucopolysaccharidosis II (MPS II) Fact Sheet for Parents

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

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

What is MPS II?

MPS II, also known as Hunter syndrome, is a rare inherited disorder that can affect many parts of the body. Individuals with MPS II cannot break down a group of complex sugars called glycosaminoglycans (GAGs) because they do not have an enzyme called iduronate-2-sulfatase (I2S). As a result, GAGs build up in cells and cause health problems.

What does an abnormal newborn screening result mean?

Newborn screening for MPS II 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 laboratory 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 II maker). Your baby's newborn screening came back suggestive for MPS II because BOTH of those tests were abnormal.

What are the symptoms of MPS II?

Individuals with MPS II can have a range of symptoms depending on how severe their disease is. Babies with the severe form of MPS II can show signs in the first year of life with progression of symptoms. In attenuated, or milder, cases of MPS II symptoms may not appear until later in childhood and progress slowly. Possible symptoms vary and can include the following:

- Developmental delays (such as delayed speech and walking)
- Large head (macrocephaly)
- Hearing loss
- Distinctive facial features, such as a prominent forehead, broad nose, and enlarged tongue
- Chronic runny nose
- Frequent upper respiratory infections
- Large belly (caused by a large liver and spleen)
- A large lump or bulge (a hernia) around the belly-button (umbilical hernia) or diaper area (inguinal hernia)
- Joint stiffness

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 II. 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 I2S 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 II treated?

The recommended treatment for MPS II is enzyme replacement therapy (ERT). ERT is a medication given weekly into your baby's vein (intravenous; IV) to replace their missing enzyme. Other treatment options may be available and will be discussed by your baby's provider. If your baby is diagnosed with MPS II, 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

- For questions about your child's newborn screening result and/or to speak with someone at the MPS Center, please call 919-228-2432.

UNC Pediatric Genetics and Metabolism

Baby's First Test

National MPS Society

Project Alive

Newborn Screening Information Center

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
N.C. DHHS is an equal opportunity employer
and provider

This fact sheet was supported by Health Resources and Services Administration (HRSA), Department of Health and Human Services (HHS) as part of a financial assistance award totaling $344,348 with 100 percent funded by HRSA/HHS. The contents are those of the author(s) and do not necessarily represent the official views of, nor an endorsement, by HRSA/HHS or the U.S. Government.