Mucopolysaccharidosis type 2 (MPS-II)

What is newborn screening?

Newborn screening (NBS) is done soon after birth to check for health conditions that can be hidden at birth. To do the screening, a nurse takes a few drops of blood from your baby's heel. This blood sample is required for all newborns.

Please remember that just because the newborn screen flags for a disorder, it does not mean that your child has a diagnosis of that disorder. It means that your child should have further testing.

KEY POINTS:

- **You have just heard that your baby might have MPS-II (Mucopolysaccharidosis type 2). Please remember that newborn screening is just a screening test. Further testing is needed before a diagnosis can be made.**
- **When MPS-II is detected early and treatment is started, many babies with the condition are able to live longer lives with improved growth, development and organ function.**

What does my baby's newborn screen show?

Your baby's newborn screen flagged for a condition called Mucopolysaccharidosis type 2, called MPS-II for short. You may also hear people refer to this condition as Hunter syndrome. The newborn screen found low levels of an enzyme called "iduronate-2-sulfatase" (called I2S for short). When the I2S enzyme is low or missing, it means that your baby could have MPS-II. Not all babies with an out-of-range screening result will go on to get a diagnosis of MPS-II. However, since some babies will be diagnosed with MPS-II, it is important to have the follow-up testing done as soon as possible, so that treatment can be started if needed. Detecting the condition early and beginning treatment may help prevent or delay some of the symptoms associated with MPS-II.

What is Mucopolysaccaridosis type 2 (MPS-II)?

MPS-II is an inherited condition that affects many different parts of the body. It is considered a lysosomal storage disorder because people with MPS II have lysosomes (the recycling center of each cell) that cannot break down large sugar molecules called glycosaminoglycans (sometimes called GAGs for short, or mucopolysaccharides). GAGs are found in the body's connective tissue. Connective tissue is found throughout the body and supports body parts and organs. Because the I2S enzyme is not working or not working well, GAGs start to build up and are stored in the body. This build-up of GAGs is what causes the variety of symptoms that have been reported with MPS-II.

MPS-II is most common in males. This is because the gene that causes MPS-II is found on the X chromosome, which is one of the sex chromosomes. However, there have also been females diagnosed with MPS-II. Diagnosing MPS-II early and beginning treatment may help prevent or delay some of the severe health outcomes associated with the condition

What health problems are seen with MPS-II?

Newborns with MPS-II often have no symptoms at birth. As children grow and develop, signs of disease may become more apparent. MPS-II is different for each person in terms of symptoms that might develop and the age at which symptoms start. People with MPS-II can have problems with how their bodies grow and work, as well as trouble with thinking and learning. They might look different than their family members. MPS-II is a lifelong condition that can result in serious health problems. However, there are treatments to help with the symptoms and slow down the disease progress. If your child is diagnosed with MPS-II, your healthcare team will discuss this treatment options in more detail.

What treatment options are available?

Some possible treatments options might include:

- Enzyme replacement therapy (ERT).
- Hematopoietic stem cell transplantation (HSCT).
- Physical Therapy (PT)
- Surgeries.

Children with MPS-II will need to be followed by a team of healthcare workers. They should see their regular doctor (pediatrician) and healthcare providers who specialize in MPS-II disease. Your baby's doctor will help arrange for a clinic visit with specialists familiar with MPS-II. The healthcare team will discuss symptoms and how to monitor and treat your child in more detail.

What if I still have questions?

We understand that this can be an overwhelming and emotional process. Many families have questions and concerns. The Connecticut Newborn Screening Network (the Network) is available to put you in touch with the best resource. To reach the Network, you can call 860-837-7870, Monday-Friday, 8:30am-4:30pm. We also recommend the website www.babysfirsttest.org as a resource.

SCAN HERE for Information about *NEWBORN SCREENING* including TIPS for preparing for blood draws and collecting urine samples on newborns;, and more

This fact sheet contains general information and is for information purposes only. Every child is different and some of this information may not apply to your child specifically. This sheet does not replace medical advice, diagnosis, or treatment from your child's healthcare providers.

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