Mucopolysaccharidosis type 1 (MPS I)

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 Mucopolysaccharidosis type 1 (MPS-I). Please understand that the newborn screening is just that: a screening test. Further testing is needed before a diagnosis can be made.**
- **When MPS-I is detected early and treatment is started, many babies with the condition are able to live longer lives with improved growth, development**

What does my baby's newborn screen show?

Your baby's newborn screen flagged for a condition called Mucopolysaccharidosis type 1 (MPS-I). The newborn screen found low levels of an enzyme called "alpha-L-iduronidase" (called IDUA for short). When IDUA is low or missing, it means that your baby could have MPS-I. Not all babies with an out-of-range screening result will go on to get a diagnosis of MPS-I. However, since some babies will be diagnosed with MPS-I, 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 might help prevent or delay some of the symptoms associated with MPS-I.

What is Mucopolysaccharidosis type 1 (MPS-I) disease?

MPS-I happens when children are missing all or some IDUA. IDUA is an enzyme needed to break down glycosaminoglycans (sometimes called GAGs for short, or mucopolysaccharides). GAGs are large sugar molecules that work in the body's connective tissue. Connective tissue is found throughout the body and supports and holds together other body parts and organs. When IDUA is not working, too many GAGs can build up in tissue throughout the body. There are different forms of MPS-I in terms of severity of symptoms: 1) severe form or 2) attenuated form. If a diagnosis is confirmed, the healthcare team can further discuss the results and what type of MPS-I your child has.

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What health problems are seen with MPS-I?

Mucopolysaccharidosis type 1 (MPS-I) disease is different for each person. People who have this disease can have problems with how their bodies grow and work, as well as trouble with thinking and learning. They might look different, too. MPS-I 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-I, your healthcare team will discuss this treatment options in more detail.

What treatment options are available?

Although MPS-I disease cannot be cured, some of the symptoms can be treated. Some possible treatments options might include:

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

Children with MPS-I 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-I disease. Your baby's doctor will help arrange for a clinic visit with specialists familiar with MPS-I. 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 Diagnosis and Treatment 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.

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 provider.

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