TEXAS Health and Human Services
Texas Department of State Health Services
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Commissioner

Newborn Screening FACT Sheet

Mucopolysaccharidosis Type 2 (MPS II)

What is MPS II?

Mucopolysaccharidosis type II (MPS II), also known as Hunter syndrome, is a rare genetic disease that affects lysosomal storage in cells of the body. Individuals with MPS II do not make enough of an enzyme (iduronate-2-sulfatase [I2S]) needed to break down sugars called glycosaminoglycans (GAGs). This causes a buildup of GAGs in the tissues that can result in problems throughout the body. The condition is caused by a change in the *IDS* gene. The affected *IDS* gene is on the X chromosome. As such, boys are usually affected because they inherit the X chromosome from their mothers who do not have symptoms of the disease, but they carry a nonworking copy of the gene. Girls get an X chromosome from their mother and father so there's less chance of getting 2 nonworking copies.

What are the symptoms of MPS II?

The timing that MPS II develops and the type of problems it causes vary between different people. Symptoms may appear as early as the first year of life or not until several years of age.

Signs of the condition may include the following:

- Large head (macrocephaly)
- Large tongue (macroglossia)
- Large liver and spleen (hepatosplenomegaly)
- Soft out-pouching around their belly button or groin (umbilical or inguinal hernia)
- Heart problems
- Stiff joints
- Sleep problems
- Hearing loss

What to expect with MPS II care:

Once a diagnosis is made, the goal is to manage the symptoms to improve quality of life and slow the progression of the disorder. Other medical specialists may also be needed depending on the symptoms the child has, such as a cardiologist and otolaryngologist (or ear nose and throat specialist). Others include:

- **Physical therapy**: It is a very important part of treating the symptoms of MPS II. Consistent physical therapy early in life can help preserve mobility and lessen pain and joint stiffness.
- **Surgery**: It might be needed to remove the tonsils and adenoids or insertion of ear drum tubes to prevent some upper respiratory infections and may reduce hearing loss. Hearing aids may be recommended for some individuals. Children may develop hydrocephaly (extra fluid around the brain structures), so a surgery to relieve the pressure inside the skull may be recommended.
- **Early treatment Enzyme Replacement Therapy (ERT)**: This can be an effective treatment for symptoms of some types of MPS II. This treatment aims to help the enzymes that are present at low levels in your baby's lysosomes. ERT may improve growth, joint movement, sleep apnea, respiratory function, pain levels, vision, and liver/spleen enlargement.

- **Hematopoietic Stem Cell Transplantation (HSCT)**: This special procedure may be recommended to improve symptoms. Hematopoietic stem cells can be found in bone marrow (the spongy tissue inside bones), the bloodstream, or the umbilical cord blood of newborn babies.
- **Clinical Trials**: Some new therapies may be available through clinical trial studies. Speak to your specialist for more details to see if your child would qualify.

Health outcomes for MPS II vary based on a child's form of the condition. For children with the mild form, early and ongoing treatment can lead to healthy growth and development. For children with the severe form, early treatment can prevent or delay some serious symptoms of the condition.

Resources:

National Organization for Rare Diseases
www.rarediseases.org/rare-diseases/mucopolysaccharidoses

National MPS Society
https://mpssociety.org/

University of Washington, Gene Reviews
https://medlineplus.gov/genetics/condition/mucopolysaccharidosis-type-ii/

NIH Lysosomal Disease
https://www.ncbi.nlm.nih.gov/books/NBK563270/

Navigate Life Texas
https://www.navigatelifetexas.org/en/diagnosis-healthcare/children-with-multiple-disabilities-rare-conditions-undiagnosed

Baby's First Test
https://babysfirsttest.org/newborn-screening/conditions/mucopolysaccharidosis-type-ii

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Where do I go for more information?
Use your phone's camera
to scan the QR code.

Next steps after a positive Newborn Screening for MPS II

Newborn screening is a blood test performed on every baby that identifies those at risk for rare disorders. Your baby's results show absent or reduced alpha-L-iduronidase (IDUA) enzyme activity. This means there is a chance that your baby may have a condition called Mucopolysaccharidosis Type 2 (MPS II). More confirmatory testing and an exam are needed as soon as possible to see if your child has MPS II.

What happens next?

- It is important to have the laboratory tests drawn. These results will help confirm or rule out the diagnosis.
- Your baby's doctor will help arrange a visit to a specialist clinic that is familiar with MPS II. The specialist will check your baby's health and discuss the test results in more detail at the visit. The team may also include a genetic counselor because this is a genetic disease. A quick diagnosis and early treatment may lessen the impact of the disease.

What is MPSII?

MPS II, also called Hunter syndrome, is a genetic disorder that primarily affects males. It interferes with the body's ability to break down and recycle specific mucopolysaccharides also known as glycosaminoglycans or GAGs. MPS II is a spectrum of disease with symptoms ranging from mild to severe. For some babies with MPS II, detecting the condition early and beginning proper treatment may help prevent or delay some of the severe health outcomes associated with the condition.

What are the symptoms of MPS II?

The severity and age when problems begin depends on which type of MPS II a person has. Bone and joint problems, enlarged liver and spleen, vision problems, and developmental delay can occur. In severe MPS II problems begin in the first or second year of life.

What is the treatment?

Early treatment involves supportive care for each organ affected and management of symptoms as needed. Treatment can include Enzyme Replacement Therapy (ERT) or stem cell transplant for those with more severe symptoms.

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