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

Newborn Screening FACT Sheet

Mucopolysaccharidosis Type 1 (MPS I)

What is MPS I?

Mucopolysaccharidosis type I (MPS I) is a lysosomal disease caused by changes (mutations) in the alpha-L-iduronidase (*IDUA*) gene. It is inherited in an autosomal recessive pattern, meaning both parents are genetic carriers of the disease but don't have the disease itself. Reduced IDUA enzyme activity causes a buildup of glycosaminoglycans (GAGs) in the lysosomes, which are parts of the cell that store and release energy. This can impact heart, respiratory, central nervous, muscle and skeletal systems. The spectrum for MPS I can range from severe to mild. MPS I is estimated to occur in about 1-3:100,000 live births.

What are the symptoms?

About 60% of the MPS I cases identified will be the most severe form, sometimes known as Hurler syndrome. Signs and symptoms are variable but may include macrocephaly or larger than typical head size, hydrocephalus or extra fluid near brain structures, heart valve abnormalities, distinctive facial features, liver and spleen enlargement, large tongue, eye problems, and bone and joint abnormalities. Delayed milestones may appear in the first year of life. This is a progressive condition and without proper treatment, children can have a shortened lifespan, sometimes living only into late childhood.

Other forms of the disease, sometimes known as Hurler-Scheie or Scheie syndrome, are less severe, also referred to as attenuated, with a later age of onset and slower progression. Symptoms may first appear any time from infancy to adulthood. Some people with the more mild type have learning disabilities, while others have no intellectual impairments.
Individuals with attenuated MPS I typically live into adulthood but may have a shortened lifespan.

What to expect with MPS I 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 signs and 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 I. 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 I. 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.

Resources:

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

National MPS Society
https://mpssociety.org/

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

MedlinePlus
https://medlineplus.gov/genetics/condition/mucopol ysaccharidosis-type-i/

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

Kennedy Ladd Foundation
https://kennedyladd.org/

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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 Screen for MPS I

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 1 MPS I. More confirmatory testing and an exam are needed as soon as possible to see if your child has MPS I.

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 I. 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 MPS I?

MPS I is a rare inherited disorder caused by non-working variants in a gene called *IDUA*. This gene controls how much IDUA enzyme the body can produce. People with MPS I are missing *IDUA* or have reduced enzyme activity so it doesn't work as well. When there is not enough IDUA enzyme activity, GAG (glycosaminoglycans) builds up in the body. This causes certain organs and muscle to not work properly.

There are different types of MPS I. The specialists will help figure out which type, if any, your child has.

What are the symptoms of MPS I?

The severity and age when problems begin depends on which type of MPS I a person has. Bone and joint problems, enlarged liver and spleen, vision problems, and developmental delay can occur. In severe MPS I, 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.