

Information Sheet

What is MPS-I?

Mucopolysaccharidosis Type I (MPS-I; also known as Hurler Syndrome) is a rare genetic condition that affects how the body breaks down complex sugars. It is part of a group of conditions called lysosomal storage disorders. Babies with MPS-I are missing or have low levels of an enzyme (a protein) called alpha-L-Iduronidase. Without this enzyme, complex carbohydrate chains (known as GAGs) build up in cells throughout the body. Over time, this buildup can affect the brain, heart, joints, and other organs.

MPS-I can be found early through newborn screening using a blood sample taken from a heel stick. If a baby's screen shows a risk for MPS-I, more testing is done using blood or urine to confirm the diagnosis. A regular doctor or a metabolic specialist will help coordinate your baby's care and arrange treatment right away.

**About 1 in
100,000**

births in the US are
affected by MPS-I.

MPS-I is treatable.

Early diagnosis through newborn screening is very important. Treatment has shown to slow or stabilize symptoms.

Symptoms

MPS-I symptoms range from severe to milder forms. Most babies look healthy at birth, but symptoms usually begin during infancy or early childhood.

Common symptoms of MPS-I include:

- Bulges around the belly button or groin (hernias)
- Frequent runny noses, ear infections, or noisy breathing
- Cloudy eyes (corneal clouding) that can affect vision
- Joint stiffness and changes in the curve of the spine
- A larger head size (macrocephaly) or distinct facial features
- Loss of skills or delays in speech and movement

Treatment

There is no cure for MPS-I, but early treatment can slow the disease and improve quality of life. Early diagnosis through newborn screening is key so treatment can start before major symptoms begin.

Treatment options include:

- **Enzyme Replacement Therapy (ERT):** Regular IV infusions that replace the missing enzyme in the body.
- **Stem Cell or Bone Marrow Transplant:** For severe MPS-I, a transplant done early in life helps the body make its own working enzyme.
- **Supportive Care:** A care team provides physical therapy, heart monitoring, and surgeries if needed to keep your child healthy and comfortable.

If left untreated, MPS-I causes life-threatening breathing problems, neurological decline, cardiac complications and shortened lifespan.

Act Quickly

Connect with a specialist when an abnormal screening result is received.

Support & Resources

Additional MPS-I Resources:

- **National MPS Society:** <https://mpssociety.org/>
- **National Organization for Rare Disorders (NORD):** <https://rarediseases.org/rare-diseases/mucopolysaccharidosis-type-i/>

Additional Newborn Screening Information:

- **Baby's First Test:** <https://www.babysfirsttest.org/newborn-screening/conditions/mucopolysaccharidosis-type-i>
- **Gene Reviews:** <https://www.ncbi.nlm.nih.gov/books/NBK1162/>
- **GARD (Genetic and Rare Diseases Information Center):** <https://rarediseases.info.nih.gov/diseases/12559/hurler-syndrome>
- **Medline Plus Condition Information for Families:** <https://medlineplus.gov/genetics/condition/mucopolysaccharidosis-type-i/>
- **HRSA Newborn Screening Clearinghouse:** <https://newbornscreening.hrsa.gov/conditions/mucopolysaccharidosis-type-i>

Kansas Department of Health and Environment Resources:

- **Kansas Newborn Screening Program:** Phone: 785-296-3363; Fax: 785-559-4245 (for reporting purposes)
- **Kansas Special Health Care Needs Program:** Care Coordination and resource assistance <https://nbss.ks.gov/shcn/>