

Information Sheet

What is MPS-II?

Mucopolysaccharidosis Type II (MPS-II), also known as Hunter syndrome, is a rare genetic condition that changes how the body recycles cell materials. Children born with MPS-II do not produce enough of an essential enzyme (a protein) called iduronate-2-sulfatase (I2S). Without enough of the I2S enzyme, complex carbohydrate chains (known as GAGs) gradually build up inside tissues and organs over time, leading to physical and developmental health issues.

MPS-II almost exclusively affects boys and can be found early through newborn screening. If your baby's screen shows a risk for MPS-II, extra blood testing will be done to confirm the diagnosis. A regular doctor and a metabolic specialist will help guide your baby's testing and care.

**About 1 in
100,000-160,000**

MALE births in the US are
affected by MPS-II.

MPS-II is treatable.

Early diagnosis through newborn screening is very important. Detecting the condition early and beginning proper treatment may help prevent or delay some of the severe health outcomes associated with the condition.

Symptoms

Babies with MPS-II usually look healthy at birth. Symptoms develop gradually over early childhood and can range from mild to severe.

Common symptoms of MPS-II include:

- Frequent ear and sinus infections, colds, or permanent runny noses
- Hernias (a soft bulge near the belly button or groin)
- Stiff joints or fingers that curve slightly inward
- Enlarged belly (caused by a large liver or spleen)
- Broad facial features that change slightly over time
- Hearing loss
- Developmental delays or loss of skills (in more severe forms)

Treatment

There is currently no cure for MPS-II, but treatments are available to manage symptoms and improve your child's life:

- **Enzyme Replacement Therapy (ERT):** Weekly IV infusions give the body the missing enzyme, helping reduce sugar buildup in organs.
- **Supportive Team Care:** A medical team will help care for your child's specific needs. This often includes physical therapy, heart care, hearing aids, and minor surgery to fix hernias.

If left untreated, MPS-II Without treatment, complex sugars continue to build up in cells throughout the body, causing progressive damage to the heart, lungs, joints, organs, and brain.

Act Quickly

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

Support & Resources

Additional MPS-II 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-ii>
- **Gene Reviews:** <https://www.ncbi.nlm.nih.gov/books/NBK1274/>
- **GARD (Genetic and Rare Disease Information Center):** <https://rarediseases.info.nih.gov/diseases/6675/mucopolysaccharidosis-mps-ii>
- **Medline Plus Condition Information for Families:** <https://medlineplus.gov/genetics/condition/mucopolysaccharidosis-type-ii/>
- **HRSA Newborn Screening Clearinghouse:** <https://newbornscreening.hrsa.gov/conditions/mucopolysaccharidosis-type-ii>

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/>