

NEWBORN SCREENING DISORDER FACT SHEET

Mucopolysaccharidosis Type-I (MPS-I)

NEWBORN SCREENING FINDINGS

A baby with low **α -L-*iduronidase* (IDUA) activity** on their newborn screening specimen(s) may have **mucopolysaccharidosis type-I (MPS-I)**. Diagnostic tests are needed to rule out or confirm MPS-I.

ABOUT THE CONDITION

MPS-I is a treatable disorder that occurs in approximately one in every 36,000 births. This condition affects the way the body breaks down large sugar molecules, called glysoaminoglycans, into smaller pieces within the body's cells. In a person with MPS-I, the IDUA enzyme that normally breaks down these sugar molecules has been altered and may not work as well or not at all. If the large sugar molecules are not broken down, they can build up in the body and lead to organ damage.

Parents who have a child with MPS-I typically each carry one altered copy of the gene involved in this process. That means, with each pregnancy, each baby will have a one in four chance of having MPS-I.

SYMPTOMS

There are different types of MPS-I that range from mild to serious. Symptoms of MPS-I may first appear around several months of age, or not until early childhood. These symptoms may include an enlarged head, lips, or tongue, as well as hearing loss, respiratory infections, joint deformities and developmental delay. If left untreated, MPS-I causes progressive multi-system disease, as well as cognitive deficits.

TREATMENT

MPS-I is treated by replacing the missing IDUA enzyme. This can be accomplished through enzyme replacement therapy and/or hematopoietic stem cell transplantation, depending on the severity of the disorder.

NEXT STEPS

The next step is to get a blood test to check the activity of the IDUA enzyme to rule out or confirm MPS-I. If the blood test is abnormal, a visit to a specialist may be needed. They may recommend additional tests or treatment.

RESOURCES

For more information about MPS-I, please see the Disorders section of our website: www.doh.wa.gov/nbs. Information can also be found at <https://www.babysfirsttest.org/>.