

Nebraska Newborn Screening Program Physician ACT sheet for REPEAT INCONCLUSIVE Mucopolysaccharidosis Type I (MPS I) Result

You Should Do The Following:

- Consult with metabolic specialist on call to determine follow up:
_____ Dr. Baker – Pgr 402-888-0404
_____ Dr. W. Rizzo – Pgr 402-888-0742
- Notify baby's family of newborn screening results. Assess status of newborn and provide follow up information as discussed with the metabolic specialist.
- Arrange for follow up as specified by the metabolic specialist

Screening Test Results

Screening for MPS I begins with measurement of the enzyme, alpha-L-iduronidase (IDUA). If the enzyme activity is decreased, a repeat newborn screen is requested. If the IDUA remains decreased on the second specimen, the lab reflexes to sequencing of the IDUA gene, which is causative for MPS I.

Condition Information

MPS I is inherited in an autosomal recessive pattern. The deficiency in IDUA activity causes an accumulation of glycosaminoglycans (GAGs) in the lysosomes. The accumulation of GAGs in the lysosomes causes the damage that results in the many symptoms patients experience.

MPS I is a disorder that affects many organ systems, has varying levels of severity and progresses at different rates. Historically the most severe type was known as Hurler disease. Patients with the much less severe presentation were classified as having Scheie syndrome. Patients with symptoms that

partially overlapped each of these were classified as having Hurler-Scheie syndrome.

Currently, MPS I might best be described as a spectrum disorder with a severe form and others that are less severely affected with varying symptoms and progression. In the most severe form symptoms begin in the first year of life. Symptoms include rapid, progressive neurocognitive impairment. Other symptoms can include skeletal abnormalities, heart and lung disease, hepatosplenomegaly, coarse facial features, hydrocephalus, hearing loss, corneal clouding, and hernias (both inguinal and umbilical). The less affected patients may have a later onset of symptoms and a slower progression of symptoms, usually without cognitive impairment.

The amount of IDUA found on the newborn screen will not predict how a patient will be affected with MPS I.

Treatment

Treatments will be determined in consultation with specialists. The specific treatments recommended are directed towards relief of the symptoms that each patient experiences. Treatments are available for all forms of MPS I.

For the most severe form, hematopoietic stem cell transplantation (HSCT) can be effective. Enzyme replacement therapy (ERT) may be given in conjunction with HSCT. Less severely affected patients are treated with ERT as well. Supportive care for specific symptoms may also be needed.