



Positive Result:

Blood Spot Screen Result Notification

Absent/Reduced Alpha-L-iduronidase (IDUA) with Elevated MPS I Biomarker

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby is missing all or some of an enzyme called alpha-L-iduronidase (IDUA) with a high amount of a specific MPS I biomarker.

What does this mean?

This means your baby may have a condition called mucopolysaccharidosis type 1 (MPS I).

What happens next?

Your baby will be referred to a specialist familiar with MPS I. They will help arrange further testing, which will give more information about if your baby has MPS I.

What is mucopolysaccharidosis type 1?

MPS I happens when children have absent or reduced amounts of an enzyme called IDUA. IDUA is needed to break down glycosaminoglycans (GAGs). GAGs are complex sugar molecules. When not broken down properly, GAGs build up in the body and can cause health problems.

There are two forms of MPS I: severe and attenuated. Newborn screening cannot tell the difference between the two forms. However, the specialists will help figure out which form, if any, your child has.

What health problems can it cause?

MPS I is a lifelong condition that may result in serious health problems. MPS I is different for each child. In severe MPS I, health problems begin in the first or second year of life. In the attenuated form, symptoms typically begin at three years of age or older, with slower progression than the severe form. If untreated, it can cause:

- Large head with specific facial features
- Bone and joint problems
- Heart problems
- Large liver and spleen
- Hernias
- Vision and hearing problems
- Sleep apnea
- Intellectual disability
- Possibility of a shortened lifespan

What treatment options are available?

MPS I can be treated. Possible treatments include enzyme replacement therapy (ERT), stem cell transplant, and supportive therapies and management like physical therapy and surgery.

Children with MPS I should see their regular doctor and doctors who specialize in MPS I.

Resources



Scan here for more information about newborn screening in Minnesota or visit our website:
www.health.state.mn.us/people/newbornscreening

MedlinePlus:

www.medlineplus.gov/genetics

Save Babies Through Screening Foundation:

www.savebabies.org

Newborn Screening Info Center:

www.newbornscreening.hrsa.gov