Family Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Absent/Reduced Alpha-L-iduronidase (IDUA) with Elevations in Dermatan Sulfate and Heparan Sulfate

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has absent/reduced alpha-L-iduronidase (IDUA) with elevations in dermatan sulfate and heparan sulfate.

What does this mean?

This means your baby likely has a condition called mucopolysaccharidosis type 1 (MPS I).

What happens next?

Your baby's doctor will help arrange for a clinic visit with specialists familiar with MPS I. The specialists will want to check on your child's health and discuss the result in more detail.

What is mucopolysaccharidosis type 1?

MPS I happens when children are missing all or some of an enzyme called alpha-L-iduronidase (IDUA). IDUA is needed to break down glycosaminoglycans (GAGs) like dermatan sulfate and heparan sulfate. GAGs are long chains of 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 different for each child. MPS I is a lifelong condition that may result in serious health problems. In severe MPS I, health problems begin in the first or second year of life. If untreated, it can cause:

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

Children with MPS I can benefit from specialized treatment.

What treatment options are available?

MPS I can be treated. Possible treatments include:

- Supportive therapies and management like physical therapy and surgery
- Enzyme replacement therapy (ERT)
- Stem cell transplant

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

Resources

Genetics Home Reference:
http://ghr.nlm.nih.gov

Save Babies Through Screening Foundation:
www.savebabies.org

Baby's First Test:
www.babysfirsttest.org

DEPARTMENT OF HEALTH

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