Family Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Absent/Reduced Acid Alpha-Glucosidase (GAA)

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has absent or reduced levels of acid alpha-glucosidase (GAA).

What does this mean?

When GAA is absent or reduced, it means that your baby likely has a condition called Pompe disease.

What happens next?

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

What is Pompe disease?

Pompe disease happens when children are missing all or some GAA. GAA is an enzyme essential in breaking down glycogen (a sugar) into glucose. When glycogen is not broken down properly, it builds up in the body and can cause health problems.

There are several types of Pompe disease: classic infantile-onset, non-classic infantile-onset, and late-onset. The severity and age when problems begin depends on which type a person has. Newborn screening cannot distinguish between the three types. However, the specialists will help figure out which type, if any, your child has.

What health problems can it cause?

Pompe disease is different for each child. Pompe disease is a lifelong condition that may result in serious health problems. If untreated, it can cause:

- Muscle weakness
- Enlarged liver
- Heart problems
- Breathing problems

Even with treatment, children with either form of infantile-onset usually have shortened lifespans.

Children with Pompe disease can benefit from specialized treatment.

What treatment options are available?

Although Pompe disease cannot be cured, some of the symptoms can be treated. Possible treatments include:

- Supportive therapies and management like physical therapy and respiratory therapy.
- Enzyme replacement therapy (ERT)

Children with Pompe disease should see their regular doctor and doctors who specialize in Pompe disease.

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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