POMPE DISEASE (GSD II)

FAMILY FACT SHEET

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 36 hours after birth. The blood is tested for rare disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called Pompe Disease, also called Glycogen Storage Disease Type II.

A positive newborn screen does not mean your baby has Pompe disease, but it does mean your baby needs more testing to know for sure.

Your baby's primary care provider or the newborn screening program will notify you to arrange for more testing.

What is Pompe disease?

Pompe disease affects an enzyme in the body that breaks down glycogen. Glycogen is a type of sugar. It is stored in structures called lysosomes. People who have Pompe disease do not make enough of the enzyme needed to break down glycogen. This makes a build up of glycogen in the lysosomes. The build up makes the lysosomes not work well. That damages the body, particularly the heart and skeletal muscles.

Pompe disease is a genetic disorder that is passed on (inherited) from parents to a child. The child's mother and father carry a gene change that can cause Pompe disease. Parents usually do not know they carry the gene change because it does not cause health problems for them.

What problems can Pompe disease cause?

Pompe disease is variable. Some people with Pompe disease show symptoms as a baby, called early-onset. They will need treatment right away. Others may not show symptoms until they are an adult, called late-onset.

A child with Pompe disease may develop:

- Heart problems
- Muscle weakness
- A large tongue
- Breathing problems
- Feeding problems

It is very important to follow the doctor's instructions for testing and needed treatment.

What is the treatment for Pompe disease?

There is no cure for Pompe disease. Treatment is life-long. Treatment can include:

- Intravenous Enzyme Replacement Therapy (called ERT)
- Physical, occupational and speech therapy
- Breathing and dietary treatment

Children who have Pompe disease should see their regular doctor. They should also see a doctor who specializes in Pompe disease as part of a special care team.

Prompt and careful treatment helps children with Pompe disease live the healthiest lives possible.

It is very important to get follow-up testing as soon as possible.

Michigan Resources and Support

Michigan Newborn Screening
Nurse Consultant
Toll-free: 1-866-673-9939
newbornscreening@michigan.gov

Michigan Metabolic Coordinating Center
Children's Hospital of Michigan 313-832-9330
University of Michigan: 1-734-764-0579

Children's Special Health Care Services
Toll-free: 1-800-359-3722

Online Resources
Medline Plus
https://medlineplus.gov/genetics/condition/pompe-disease/
National Organization for Rare Diseases
www.rarediseases.org/pompe

MDHHS Michigan Department of Health & Human Services | Michigan Newborn Screening

Phone 1-866-673-9939
www.michigan.gov/newbornscreening
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