Newborn Screening and Pompe Disease
A Parent's Guide

Indiana Department of Health

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What is newborn screening?

Soon after birth, babies have a small amount of blood taken from their heel. The blood is used to test for some rare but treatable health conditions. This test is part of the Indiana Newborn Screening Program.

Your baby's newborn screen was positive for a condition called Pompe disease (also called glycogen storage disease, type 2). This does **not** mean your baby has Pompe disease, but more testing is needed for diagnosis. While you wait for more test results, this sheet can help you learn more about Pompe disease.

What is Pompe disease?

Pompe disease is a condition at birth. It affects how babies break down a kind of sugar called glycogen.

Most people have a special helper in their body called an enzyme that breaks down glycogen. This enzyme is called acid alpha glucosidase, or GAA. Babies with Pompe disease do not have enough of this enzyme. That means that glycogen can build up in their bodies, especially in the heart and muscles. Too much glycogen can cause health problems if not treated.

There are two types of Pompe disease: infantile-onset Pompe disease (also called IOPD) and late-onset Pompe disease (also called LOPD).

Spot the Signs

Every baby with Pompe disease is different. Not all babies will have symptoms right away. The symptoms depend on the type of Pompe disease.

IOPD starts early in life and symptoms may include:

- Heart problems
- Weak muscles
- Floppy arms and legs
- Trouble feeding and growing
- Trouble breathing

LOPD often appears later in childhood or adulthood. It may cause milder symptoms of weak muscles and trouble breathing.

What causes Pompe disease?

Pompe disease is passed from parents to their baby. A baby with Pompe disease gets one changed GAA gene from each parent. These changes stop the body from making enough of the GAA enzyme.

Babies who only have one changed GAA gene do **not** have Pompe disease.

What is the treatment for Pompe disease?

There is no cure for Pompe disease, but enzyme replacement therapy (ERT) can help with symptoms. ERT gives the body the GAA enzyme it is missing. The medicine is given through an IV once a week or once every other week.

For babies with IOPD, starting ERT as early as possible – within the first month – is very important. It can help your baby live longer and protects the heart, muscles, and motor skills.

For children with LOPD, starting treatment depends on symptoms. If your baby has LOPD but no symptoms, doctors may recommend close monitoring before starting ERT. This helps make sure children start treatment at the right time and avoid medicine they do not yet need.

Your baby may also need other care to stay healthy, such as:

- Physical therapy
- Breathing support
- Help with feeding and nutrition
- Ways to avoid infections

Your baby will also have regular doctor visits to check their heart, muscles, and lungs. Most children will need some kind of treatment or support for their entire life.

What happens next?

For now, take care of your baby as usual. If the test results are positive again for Pompe disease, your baby's doctor will explain what to do next.

Babies with Pompe disease will see a metabolic geneticist – a doctor who helps care for babies with Pompe disease – and will also keep seeing their regular doctor (pediatrician). If needed, your baby may be referred to other doctors, too. Your baby's doctors will work as a team to help with the tests and treatment your baby needs.

Thanks to newborn screening, babies with Pompe disease can get early care and live a healthier life.

Where are Indiana's metabolic genetics clinics?

Indiana's metabolic genetics clinics are located at Riley Hospital for Children at IU Health in Indianapolis (317-274-3966) and The Community Health Clinic in Shipshewana (260-593-0108).

baby's first test

NORD® National Organization for Rare Disorders

For additional information, visit:
rarediseases.org/rare-diseases/pompe-disease/

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