

Family action sheet for Pompe disease

If a newborn blood screening result shows that your baby could have a serious condition called Pompe disease, this result does not mean that your baby has this condition, but more tests are needed.

All children born in California have a routine blood screen shortly after birth. The goal of this newborn screening is to find those at risk for serious medical conditions. Babies can look healthy at birth and still have one of these conditions. Babies with these conditions benefit from early diagnosis and treatment.

What is Pompe Disease?

Pompe disease prevents the body from breaking down sugars properly. This can affect many parts of the body, leading to problems with the heart, lungs, and muscles, as well as difficulties feeding and gaining weight.

There are two major forms of Pompe disease. The most severe form of Pompe occurs in the first few months of life and requires immediate treatment. The less severe form can present at any age and may not require treatment right away.

Pompe is an inherited condition. This means it is passed from parents to children.

Is there treatment for Pompe disease?

Babies with Pompe disease are treated with enzyme replacement therapy (ERT) to help with symptoms. Other supportive care may include respiratory, physical, and nutritional therapies.

Newborn screening takes blood from a newborn's heel to screen for medical conditions



Next steps

1. Your baby's medical provider will refer you to specialists with Pompe disease experience.

The specialists will arrange for further testing and evaluation. More tests are needed to find what treatment or monitoring will be needed for your baby.

2. Work closely with your baby's medical provider and specialists and follow their recommendations.

It is important that you keep all appointments and start treatment if instructed.

Where can I get more information?

Your baby's primary care doctor or Pompe specialist is the best person to teach you about Pompe. Find more information at these websites:

• [Baby's First Test](https://www.babysfirsttest.org)

(<https://www.babysfirsttest.org>). Type "Pompe" into the "Find a Condition" Box. Follow the link. For Spanish, click on the top right black box marked "en Español."

• [Health Resources & Services Administration \(HRSA\)](https://newbornscreening.hrsa.gov/conditions/pompe-disease)

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