



### Newborn Screening & Pompe Disease

In the U.S., newborns are tested at 24-48 hours after birth for a number of serious medical conditions. Newborn screening helps babies have healthier lives. It can save lives.

When a newborn screening test result is not normal (out-of-range), the baby's doctor or state newborn screening program contacts the baby's family. Many states test for Pompe disease. Other states plan to start testing.

Learn more about newborn screening for Pompe disease:



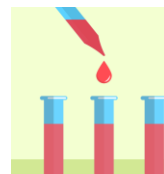
- HRSA-Pompe  
<https://newbornscreening.hrsa.gov/conditions/pompe-disease>
- EveryLife Foundation for Rare Diseases  
<https://everylifefoundation.org/newborn-screening-take-action/pompe-disease/>
- Search which states test for Pompe  
<https://www.babysfirsttest.org/>

### Follow-up with Your Baby's Doctor

If your baby's newborn screening test for Pompe disease was not normal (out-of-range), take your baby to the doctor as soon as possible.

An out-of-range newborn screening test does not mean that your baby absolutely has Pompe disease. Your baby's doctor will do more tests to confirm the diagnosis. Your baby may need:

- a heart exam with chest x-ray or echocardiogram
- blood and urine tests
- a genetic test



Tests will show if your baby has Pompe disease. Early treatment helps prevent harmful effects that can happen soon after birth.

Learn more about testing and follow-up care:

- Baby's First Test  
<https://www.babysfirsttest.org/newborn-screening/conditions/pompe>
- Acid Maltase Deficiency Association  
<https://amda-pompe.org/treatingpompe>

### If Your Baby Has Pompe Disease

Finding out that your baby has Pompe disease can be hard. You may feel scared, sad, confused, overwhelmed. Whatever you are feeling is understandable. What can you do now?

The Acid Maltase Deficiency Association (AMDA) is a non-profit patient support organization. We can provide you and your family with the information, support, and resources you need to move forward. We are here to help you and your family as you care for your baby. Please reach out to AMDA's Patient Advocate, Marsha Zimmerman, RN, at [marsha.zimmerman@amda-pompe.org](mailto:marsha.zimmerman@amda-pompe.org)

One of the best next steps is to learn all you can about Pompe disease. We suggest reviewing the parent materials and other resources on the AMDA website: <https://amda-pompe.org>

Be sure to read the University of Michigan's *Pompe Disease Family Education Booklet*.  
<https://www.med.umich.edu/1libr/Pediatrics/Genetics/PompeDiseaseBooklet.pdf>