

# Positive Result:

## Blood Spot Screen Result Notification



### Immunoreactive Trypsinogen (IRT) $\geq$ 120 ng/mL with No CFTR Variants Identified

#### What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has a very high IRT level. IRT is a protein made by the pancreas. IRT can be elevated for a number of reasons, including cystic fibrosis (CF). Because the IRT was elevated, your baby's blood spots were tested for the 39 most common changes (variants) to the gene that causes CF (CFTR gene). Your baby did not have any of those most common changes.

#### What does this mean?

Most likely the IRT level is very high because some of your baby's meconium (first bowel movement) got on the newborn screening card or because your baby was stressed at birth.

Your baby likely does not have CF; however, it is possible that your child has changes to the cystic fibrosis gene that newborn screening did not find because we only test for the most common changes.

#### What happens next?

Your baby's doctor will collect a repeat newborn screen. Most repeat screens return with normal results and no other testing is needed. If the IRT remains elevated, your baby's doctor will arrange for more testing at a cystic fibrosis center with specialists familiar with CF.

#### What is cystic fibrosis?

CF is a disease that causes thick, sticky mucus to build up. This mucus can lead to problems with breathing and lung infections. This mucus can also make it harder for the body to break down food.

#### What health problems can it cause?

CF is different for each child. CF is a lifelong disease that may result in serious health problems. Children with CF can develop:

- Poor weight gain
- Greasy or oily bowel movements
- Poor growth
- Coughing and wheezing
- Lung infections

Children with CF can benefit from prompt and careful treatment.

#### What treatment options are available?

Although CF cannot be cured, the symptoms can be treated. Possible treatments can include:

- Prescription enzymes to help absorb food better
- Healthy, high-calorie diet
- Vitamins
- Medications to prevent infections and help with breathing
- Ways to help clear mucus from the lungs

Children with CF should see their regular doctor and a doctor who specializes in CF.

#### Resources

Genetics Home Reference:  
<http://ghr.nlm.nih.gov>

Save Babies Through Screening Foundation:  
[www.savebabies.org](http://www.savebabies.org)

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