

# Positive Result:

## Blood Spot Screen Result Notification



### Elevated Immunoreactive Trypsinogen (IRT) and 2 CFTR Variants Identified (Both CF-Causing)

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

The newborn screen that was collected at birth found that your baby has a 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 was found to have two disease-causing changes in the cystic fibrosis gene.

#### What does this mean?

It is very likely that your baby has CF.

#### What happens next?

Your baby's doctor will help arrange for more testing at a cystic fibrosis center with specialists familiar with CF. The specialists will want to see your child as soon as possible to start treatment. They will arrange for a sweat test to confirm your child has 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)