Positive Result:
Blood Spot Screen Result Notification

Immunoreactive Trypsinogen (IRT) ≥ 100 ng/mL
with 1 CFTR Variant 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 was found to have one change in the cystic fibrosis gene.

What does this mean?
When only one change in the cystic fibrosis gene is found, it is most likely that your baby is a carrier for CF (unaffected). Two changes in the cystic fibrosis gene are needed to cause CF, so it is less likely that your baby has cystic fibrosis (affected). Since newborn screening does not test for every change in the cystic fibrosis gene, it is still possible that your child has a second change in their cystic fibrosis gene that would cause CF. There is more concern for a second change in the cystic fibrosis gene when there is a very high IRT level.

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.

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 do a sweat test to find out if your child has CF. You will be able to speak with a genetic counselor about the meaning of the results for your family.

Resources
Save Babies Through Screening Foundation: www.savebabies.org
Baby's First Test: www.babysfirsttest.org

Newborn Screening Program, 601 Robert St. N., St. Paul, MN 55155
Phone (800) 664-7772, Fax (651) 215-6285 *interpreters available
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