NORTH CAROLINA NEWBORN SCREENING PROGRAM

Elevated IRT and 1 CFTR Variant Fact Sheet for Parents

What was found on your newborn's screen?

The newborn screen that was collected at birth found that your baby has a high IRT level. IRT, which stands for "immunoreactive trypsinogen", is a protein made by the pancreas. IRT can be elevated for several reasons, one of which is cystic fibrosis (CF). Because the IRT was elevated, your baby's blood spots were tested for the 139 most common changes (variants) to the gene that cause CF (CFTR gene). Your baby was found to have one change in the cystic fibrosis gene.

What is cystic fibrosis (CF)?

CF is a genetic 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.

Children with CF can develop:

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

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.

What happens next?

Your baby's doctor will help arrange for more testing at a cystic fibrosis center with CF specialists familiar with CF. The specialists will do a sweat test to find out if your child has CF. The sweat test will likely be scheduled for when your baby is between 2 and 4 weeks old. You will be able to speak with a nurse or genetic counselor about the meaning of the results for your family.

How is this condition managed?

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
- Therapies to help clear mucus from the lungs

Children with CF should see their regular doctor and a doctor who specializes in CF at an accredited CF center.

Where do I go for more information?

Use your phone's camera to scan the QR code below.

Cystic Fibrosis Foundation:
https://www.cff.org/

NC DEPARTMENT OF
HEALTH AND HUMAN SERVICES

State of North Carolina Department of Health and Human Services
www.ncdhhs.gov
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