

Positive Result:

Blood Spot Screen Result Notification



Elevated Immunoreactive Trypsinogen (IRT) and 2 CFTR Variants (One or Both of Varying Clinical Consequence) Identified

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 changes in the cystic fibrosis gene.

What does this mean?

Your baby likely has cystic fibrosis transmembrane conductance regulator (CFTR)-related metabolic syndrome (CRMS).

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 CRMS or CF.

What is CRMS?

CRMS is a milder variation of cystic fibrosis (CF). This diagnosis is often given to people who have higher than normal IRT levels but not as high as those with classic CF. People with CRMS can develop some health problems that are similar to those with CF.

What health problems can it cause?

CRMS is different for each child. Some children can develop problems with:

- Breathing
- Intestines
- Pancreas
- Reproductive tract

Many children never develop symptoms.

What treatment options are available?

There is no cure for CRMS. Routine treatment is not needed unless symptoms develop.

Children with CRMS should see their regular doctor and a doctor who specializes in CF. These regular checkups will help to find and treat any symptoms early if they were to occur.

Resources

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

Save Babies Through Screening Foundation:
www.savebabies.org

Baby's First Test:
www.babysfirsttest.org