

CFTR-Related Metabolic Syndrome (CRMS)

Your child has CFTR-Related Metabolic Syndrome (CRMS). Your infant had a newborn screen test for cystic fibrosis (CF) that gave an intermediate result. Your child does not have CF, but there are reasons why we want your child to have regular check-ups with a doctor who is a cystic fibrosis specialist. To understand CRMS, you need to know a little bit about CF.

What causes CF?

CF is a genetic disease. Genes are what tells your body if you will have blue eyes or you will have curly hair. Genes come in pairs. You get one from your mother and one from your father.

When there is a change in the code in a gene it's called a mutation. Some mutations don't cause any problems, but some can cause problems like CF. People with CF have a disease-causing mutation that they got from both their mother and father.

What are the symptoms of CF?

- Salty skin
- This and sticky mucus buildup in certain parts of the body:
 - Breathing tubes
 - Sinuses
 - Intestines
 - Organs that connect to intestines (like pancreas)
 - Reproductive tract

Often, people get a germ called *Pseudomonas* (pronounced “soo-dah-MOAN-us”). When we find this germ, we treat it because it can worsen lung function. People with CF can get very serious and permanent lung problems.

What is CRMS?

We say that a child has CRMS when they have had a sweat test or a genetic test that gives an intermediate result. Sweat tests are used to diagnose cystic fibrosis (CF).

Your child does not have CF, but we found your child has CRMS because either:

- the amount of salt in your child's sweat is higher than most children (though not high enough to say that he has CF), or
- your child has one or two mutations in her CF genes.

What does this mean?

This result means your child has a higher risk of problems in:

- Breathing tubes or sinuses
- Intestines and organs that connect to the intestines (like the pancreas)
- Reproductive tract

What are the symptoms of CRMS?

Your child is likely to remain healthy, though we cannot clearly predict his future health. Though some people with CRMS have dealt with the problems listed above, we don't know how many **do not** have those problems. We think that it is best for your child to have regular check-ups with a CF specialist so that we can find and treat any changes early **if** they happen.

You should see your regular doctor and maybe your CF specialist if your child:

- Is not gaining weight.
- Has loose stools, very bad gassiness or constipation that last more than 2 weeks.
- Has very bad stomach aches.

- Has coughing or wheezing that last more than 2 weeks.

If your CF specialist sees your child in an office with people with CF, they may take special precautions to be sure that your child is not exposed to the *Pseudomonas* germ. You should know this germ is everywhere, and even healthy babies might have it.

What can we do to keep our child healthy?

Your child should not be exposed to cigarette smoke. All children who are over 6 months of age should receive the yearly flu shot.

Your health care team may have given you this information as part of your care. If so, please use it and call if you have any questions. If this information was not given to you as part of your care, please check with your doctor. This is not medical advice. This is not to be used for diagnosis or treatment of any medical condition. Because each person's health needs are different, you should talk with your doctor or others on your health care team when using this information. If you have an emergency, please call 911. Copyright © 4/2019 University of Wisconsin Hospitals and Clinics Authority. All rights reserved. Produced by the Department of Nursing. HF#6990.