CFTR-Related Metabolic Syndrome (CRMS)

(CFTR stands for Cystic Fibrosis Transmembrane Regulator protein, which is the salt channel that the “CF gene” makes; see “What Causes CF”)

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

What is CRMS?

We say that a child has CFTR-Related Metabolic Syndrome (CRMS) when they have had a sweat test or a genetic test that gives an intermediate result. Sweat tests are used to make the diagnosis of cystic fibrosis (CF), a genetic (inherited) disease. CF causes thick mucus to get stuck in the breathing tubes or sinuses, the intestines and organs like the pancreas that are connected to the intestines, or the reproductive tract. Your child does not have CF, but one of two things makes us say that he or she has CRMS:

- The amount of salt in your child’s sweat is higher than most children, although it is not high enough for us to say that he or she has CF. This could mean that your child is at higher risk to have problems in the breathing tubes or sinuses, the intestines and organs like the pancreas that are connected to the intestines, or the reproductive tract.

- Your child has one or two mutations in his or her CF genes that don’t cause CF, but could mean that your child is at higher risk to have problems in the breathing tubes or sinuses, the intestines and organs like the pancreas that are connected to the intestines, or the reproductive tract.

What causes cystic fibrosis?

To understand CRMS you need to know something about cystic fibrosis (CF). CF is a genetic (inherited) disease. Genes are what tells our body things like “you will have blue eyes” or “you will have curly hair”. Genes comes 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 at all, but some can cause diseases like CF. People with CF have a disease-causing mutation in each of their two CFTR genes, so they inherited one mutation from their mother and one mutation from their father.
What are the symptoms of cystic fibrosis?

The CFTR gene controls the salt channels in skin, and because they have mutations that cause disease, people with CF have very salty skin. The CF gene also controls the salt channels in parts of the body that are lined with tissues that are like skin – the breathing tubes and sinuses, the intestines and organs like the pancreas that are connected to the intestines, and the reproductive tract. People with CF get thick and sticky mucus in these parts of the body. The breathing tubes get clogged with thick mucus and often people get a germ called Pseudomonas (pronounced “soo-dah-MOAN-us”). When we find Pseudomonas, we treat it because people who have this germ can have worse lung function. People with CF can get very serious and permanent lung problems.

What are the symptoms of CRMS?

We cannot clearly predict the future health of your child, although he or she is likely to remain healthy. Some people with CRMS have developed problems in the breathing tubes or sinuses, the intestines and organs like the pancreas that are connected to the intestines, or the reproductive tract, but we don’t know how many people with CRMS don’t develop these problems. We think that the best thing to do is for your child to have regular check-ups with a CF specialist so that we can find and treat any early changes if they happen.

You should see your regular doctor and possibly 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 where there are people with CF, they may take special precautions to be sure that your child is not exposed to the germ called Pseudomonas. It is important to know that Pseudomonas is everywhere, and even healthy babies might have Pseudomonas.

What can we do to keep our child healthy?

As is true for all children, people with CRMS should not be exposed to cigarette smoke. All children who are over 6 months of age should receive yearly influenza vaccine.

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