



Cystic Fibrosis Canada

Your Child and Cystic Fibrosis



“When I was first told that Sophia had cystic fibrosis, I was devastated. I went through a mourning process. I cried a lot. Then I started to change the way I thought about life. I decided that Sophia and I were going to live each day as it came. We would try and make at least one happy memory every day, and we would do everything we could to keep her healthy and happy.”

Katie Schulz, Dundas, Ontario

What is cystic fibrosis?

Cystic fibrosis (CF) is an inherited disease affecting approximately one in every 3,600 Canadians. Inherited means that cystic fibrosis is passed down from parents to children through genes. In order to have cystic fibrosis, a child must inherit two CF genes (one CF gene from each parent). In most cases, the parents themselves do not have cystic fibrosis.

Cystic fibrosis affects mainly the lungs and digestive system, but cystic fibrosis also affects the sinuses, liver, pancreas, and reproductive (sex) organs. Cystic fibrosis is caused when a protein in the cells within the body does not work properly. This protein, called the Cystic Fibrosis Transmembrane Regulator protein (CFTR), works to move salt and water in and out of cells in the body. When the CFTR protein is not working properly, it causes mucus in the body to become thicker and stickier than normal. Mucus is normally a thin, slippery material made by the body to

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line the insides of tubes in the body (for example the airway tubes in the lungs and the tubes or bowels of the digestive and reproductive systems). When mucus becomes thick and sticky it can block or plug some of the tubes in the body.

In the lungs, a build-up of thick or sticky mucus causes breathing problems. The most common problem is coughing and an increase in sputum (also known as phlegm or spit) from the lungs. Thick mucus in the lungs also makes it easier for bacteria to grow, which causes infections in the chest. Over time, these infections can cause damage and scarring in the lungs. When mucus builds up in the digestive tract, it makes it difficult to digest and absorb nutrients from food. In most people with cystic fibrosis, the small tubes within the pancreas become blocked and damaged. When this happens, digestive enzymes made by the pancreas can no longer reach the intestine.

Because treatments have helped to improve nutrition and absorption of food, most deaths related to cystic fibrosis are because of lung disease.

Approximately 4,000 Canadian children, adolescents, and adults with cystic fibrosis attend specialized CF clinics. Currently, no cure exists. Every day, research is being done to improve the treatment of cystic fibrosis with the hope of one day finding a cure or effective control.

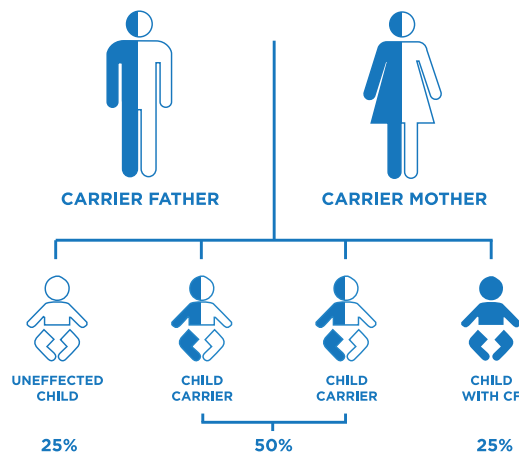
What causes cystic fibrosis?

Cystic fibrosis is a genetic inherited disease that individuals are born with. Cystic fibrosis occurs when a child inherits two abnormal genes, one from each parent.

Approximately one in every 25 Canadiansⁱ is a CF carrier, meaning they have one abnormal version of the gene responsible for cystic fibrosis. Carriers do not have, and can never get, cystic fibrosis. In most cases, they are not even aware they are carriers.

Each time two individuals who are carriers have a child together, there is a:

- ◆ 25 percent chance the child will be born with cystic fibrosis (they will have two abnormal CF genes)
- ◆ 50 percent chance the child will not have cystic fibrosis, but will be a carrier (they will have one abnormal CF gene, and one normal gene)
- ◆ 25 percent chance the child will not have cystic fibrosis and will not be a carrier (they will have two normal genes)



Each pregnancy carries the same risk of a child inheriting an abnormal gene. For instance, two parents who are carriers may have several children with cystic fibrosis, or none at all. Carrier status (that is, finding out whether you or your partner has a CF gene) can be determined through genetic testing. If you, your partner, or child is a carrier, genetic counselling is available.

ⁱ CF carrier rates vary by ethnicity.

What are the symptoms of cystic fibrosis?

Cystic fibrosis affects many parts of the body. The most common symptoms include:

- ◆ persistent cough with production of thick mucus (phlegm or spit from the lungs)
- ◆ wheezing and shortness of breath
- ◆ frequent chest infections which may include pneumonia and bronchitis
- ◆ infections in the sinuses (sinusitis)
- ◆ bowel and digestive problems, such as bowel blockage or constipation
- ◆ large, smelly, oily or greasy stools (bowel movements) because of difficulty in absorbing fat from food
- ◆ weight loss or failure to gain weight, despite possible increase in appetite
- ◆ salty tasting sweat (this happens because the CFTR protein in the skin's sweat glands does not carry salt across the sweat gland normally)
- ◆ infertility (in males, because the tube which carries sperm out of the body, the vas deferens, does not develop), or decreased fertility (in females, because the mucus of the fallopian tubes, uterus and cervix is abnormally thick)

Cystic fibrosis is not always immediately recognized in young children. The symptoms of cystic fibrosis may not be very obvious at first, or the symptoms are thought to be due to other causes.

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One symptom of cystic fibrosis that is easily identifiable within the first few days of life is *meconium ileus*. *Meconium ileus* is an intestinal (bowel) blockage in a newborn baby.

It is diagnosed when the baby cannot have a bowel movement because the bowels are blocked by putty-like material. *Meconium ileus* can sometimes be resolved with medical therapy such as washing out the bowel with enemas; however, in more serious cases, surgery may be required to remove the blockage.

How is cystic fibrosis diagnosed?

In many provinces, early diagnosis of cystic fibrosis happens through newborn screening programs. Shortly after a baby is born, a small blood sample is taken from the baby's heel. The blood sample is sent to a laboratory, where it is screened for a number of different health conditions. The laboratory screens for cystic fibrosis by testing for a chemical made by the pancreas called Immunoreactive trypsinogen (IRT). If the initial IRT screen is high or positive, more testing at a specialized CF clinic is needed to determine if the baby has cystic fibrosis or not. These tests will include a sweat test, but other tests may also be done such as detailed genetic testing looking for CF genes. If more genetic testing is needed, a cheek swab or a blood sample may be required.

The sweat test is a good test for diagnosing cystic fibrosis. It is an accurate test, detecting abnormal function of the cystic fibrosis protein. It measures the amount of chloride (salt) present in the sweat. Cystic fibrosis is the most common cause of an elevated sweat chloride level. The sweat test does not hurt at all, and is done by collecting a small amount of sweat from the skin.

In children who are not diagnosed by newborn screening, most are diagnosed with cystic fibrosis once they start to show symptoms. The most common of these symptoms include chronic coughing or not gaining weight as well as expected. There may also be signs of bowel movements which are not normal. These symptoms will signal to your doctor to order a sweat test.

How is cystic fibrosis treated?

Although there is currently no cure for cystic fibrosis, there are excellent treatments that are improving the lives of people living with this disease. Canadians with cystic fibrosis are living healthier, longer lives than ever

Today, nearly 60 percent of Canadians with cystic fibrosis are adults.

before. In 1960, most children with cystic fibrosis did not live long enough to attend kindergarten. Today, nearly 60 percent of Canadians with cystic fibrosis are adults.

The degree of involvement of cystic fibrosis varies from person to person therefore each child with cystic fibrosis will be prescribed a personalized treatment. Treatments should only be changed in consultation with the CF clinic team.

Pulmonary

An important part of CF care is treating and preventing progression of lung disease. Clearing mucus from the airways in the lungs is done with chest physiotherapy which is also known as airway clearance. A variety of ways can be used, including clapping the chest, breathing exercises, and vibrating vest machines.



Special inhaled solutions may be used to thin and loosen the mucus in the lungs, while other medications called bronchodilators help open the airways to improve mucus clearance.

To treat lung infections, your physician will often prescribe oral or inhaled antibiotics.

Gastrointestinal

Approximately 85 percent of people with cystic fibrosis experience digestive problems. These problems include difficulty in absorbing the fat, protein, and carbohydrates from food. Most people with cystic fibrosis need special pancreatic enzymes, a special diet with increased calories and protein, and extra vitamins including vitamins A, D, E, and K.

Pancreatic enzymes, which are taken by mouth, help the body absorb nutrients from food, and reduce both the number and size of stools (bowel movements), and the amount of flatulence (gas), abdominal pain, and distension (bloating).

People with cystic fibrosis also usually need to increase salt intake to replace the extra amount of salt which gets lost in sweat.

Approximately 15 percent of people with cystic fibrosis have a working pancreas (this is known as pancreatic sufficiency). In most cases, these people do not need large doses of pancreatic enzymes, and in some cases may not need enzymes at all. However, sometimes in these children, the pancreas can become irritated or inflamed (called pancreatitis) which requires special attention and treatment. Your doctor will let you know if this is the case for your child.

A small number of people with cystic fibrosis can develop liver disease, as the small tubes inside the liver can also become irritated and damaged.

The team approach to care

A team of healthcare professionals play an important part in your child's care. Depending on the CF clinic team, it will often include a nurse practitioner, social worker, dietician, pharmacist, physiotherapist, and respiratory therapist, in addition to the physician and nurse coordinator.

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The physician

Often your CF care team will be comprised of several physicians, who will rotate

in seeing each patient in clinic. The physicians will make all CF medical decisions in consultation with the team, prescribe required medications and lead the team in care provided. Often these physicians will be responsible for diagnosing and admitting patients to hospital when necessary. Many of the physicians are also involved with CF research such as clinical trials for new CF treatments.

The nurse

The nurse(s) will be your main contact when health issues arise. They get to know each CF patient and their medical issues, and can provide answers to your question and/or refer to another team member. The nurse(s) will see each patient and their family during a clinic visit and often visit them if there is a hospital admission.

The physiotherapist and respiratory therapist

The physiotherapist will teach you how to perform various airway clearance techniques (chest physiotherapy) and perform these treatments when admission to hospital is necessary. Airway clearance techniques are customized to each patient and are reviewed in clinic by your therapist.

The respiratory therapist is responsible for helping with your child's nebulizers, compressors, and all other equipment you may need for your child's treatments. In hospital, the respiratory therapist will set up and maintain equipment for aerosol (inhaled) therapy. The therapist may supervise treatments and perform pulmonary function tests (breathing tests) as required.

The dietician

The dietician will review your child's diet, and give advice about providing your child with the right foods to gain and maintain weight. Food and good nutrition are vital for patients with cystic fibrosis. The dietician will also teach patients about the use of enzymes and when to take them.

The social worker

The social worker helps patients with CF and their families cope with cystic fibrosis. They assist in the search for community resources such as financial assistance, tax credits and educational opportunities. The social worker can provide support with emotional difficulties and refer patients to a psychologist or psychiatrist for further support if needed.

The pharmacist

The pharmacist supplies CF medication and is available to explain the purpose of each drug, the dosage and best routine for giving each medication. The pharmacist is a great resource to help with accessing information about provincial drug coverage and, along with your social worker, will help with special access forms if they are needed to gain access to medications.

Should I take special precautions with my child?



With excellent attention to treatment and regular follow-up, most children with cystic fibrosis now lead relatively normal lives. Children with cystic fibrosis often have excellent school attendance records and most are able to participate in games and sports. Physical activity is

encouraged, since it helps children maintain excellent physical fitness and lung health. If you have questions, ask your physician how much your child should be permitted to do.

Like any child, your child should avoid unnecessary contact with anyone known to have a cold or contagious illness. He or she should be given the usual protective immunizations against childhood diseases.

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According to medical science, people with cystic fibrosis should avoid being in close contact with each other in order to control the

transmission of dangerous infections and to protect their own health. An infection prevention and control policy is available at www.cysticfibrosis.ca.

Infection Control and Prevention

How can I connect with other families affected by cystic fibrosis?

Please see our webpage at: www.cysticfibrosis.ca to read our infection Control Guideline for the protection of you and your child.

Join the My CF Canada Network, Canada's first-ever social network designed by CF patients specifically for CF patients to connect and share important information about living with this severe and potentially fatal genetic disease. Canadians with cystic fibrosis can discuss the treatments, programs and services available in their province/region, via video chat and instant message, share stories and personal issues, and receive support for advocacy and fundraising initiatives from a much broader community.

Cystic Fibrosis Canada acknowledges Novartis Pharmaceuticals Canada Inc. for the collaboration that has made the My CF Canada Network possible.

Sign-up today at www.mycfnetwork.com

For more information about cystic fibrosis, visit cysticfibrosis.ca

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www.cysticfibrosis.ca

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