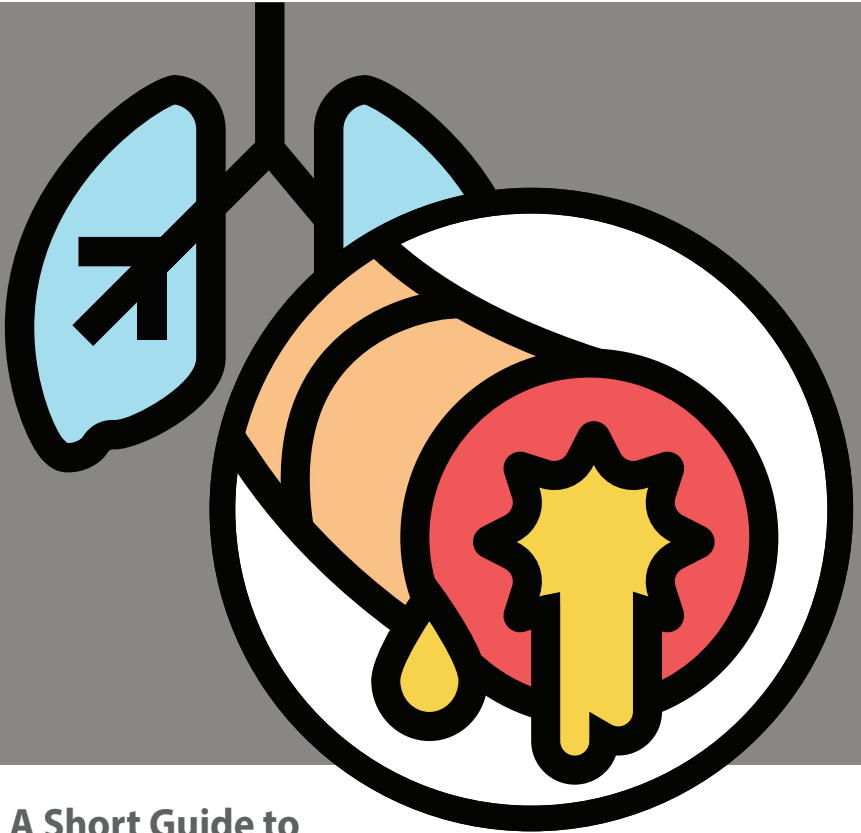




Montreal Chest Institute

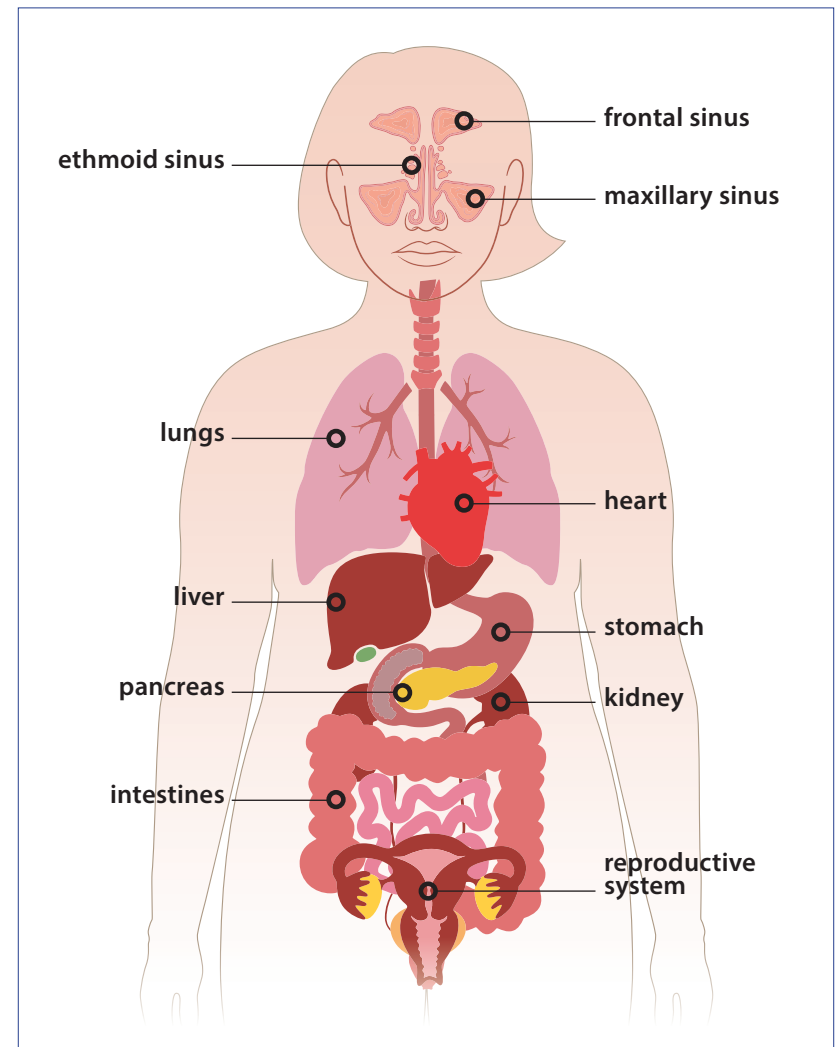
A Rare Lung Disease //



A Short Guide to
Cystic fibrosis (CF)

// What is cystic fibrosis?

Cystic fibrosis (CF) is a common genetic disease that you inherit from both biological parents. About 1 in 3,600 Canadians are born with CF. It is an autosomal recessive disease, meaning that you must inherit a mutation from each parent. If you have only 1 mutation, you generally have no symptoms, you don't have CF, you are simply considered a "carrier" for the mutation.



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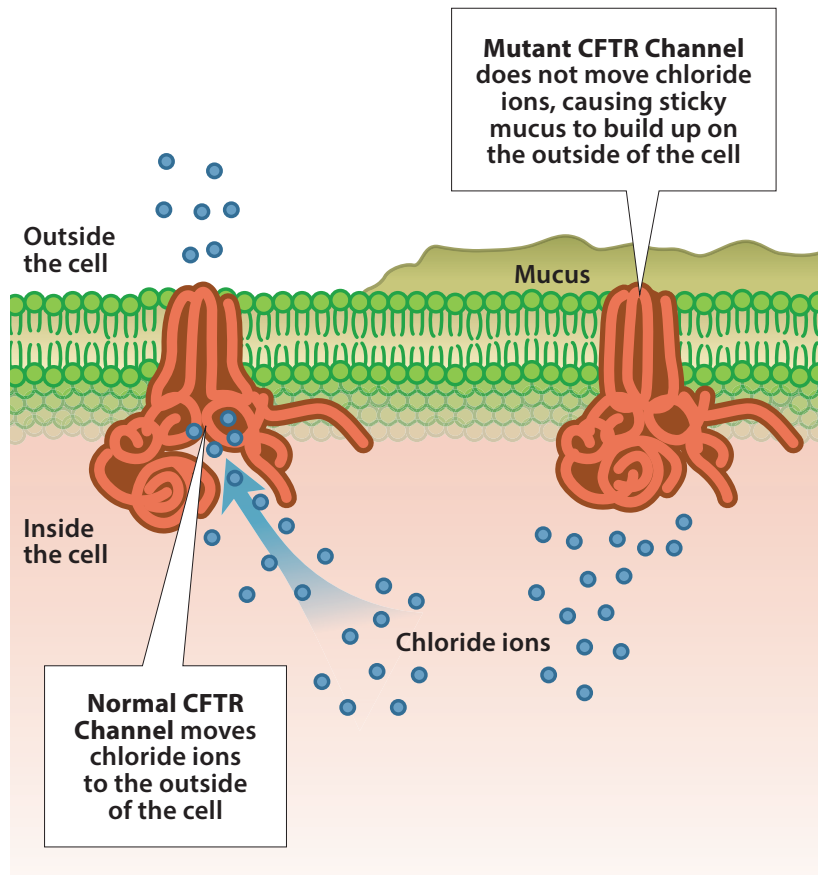
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// What causes CF?

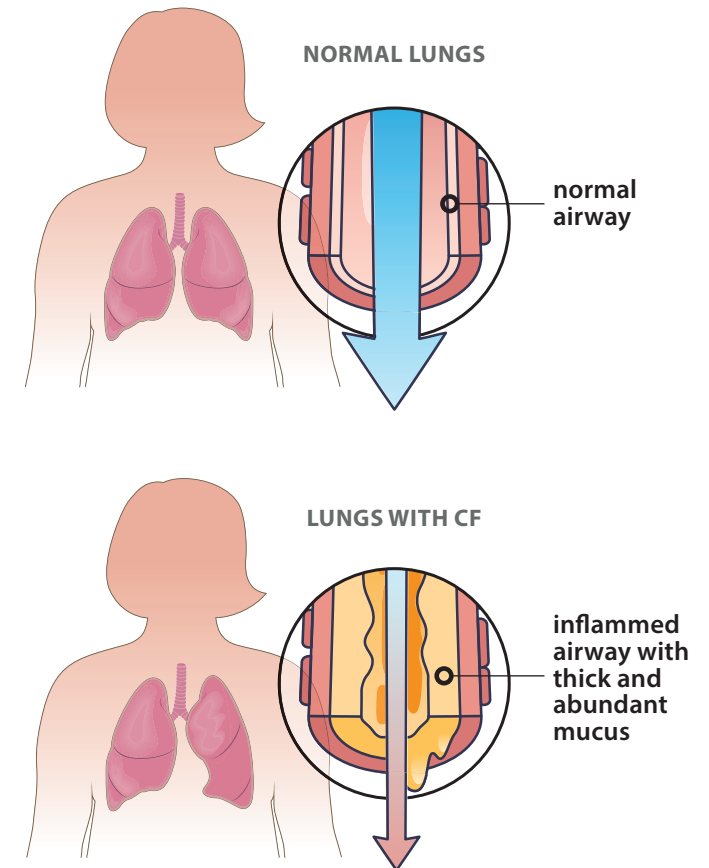
CF is caused by a dysfunction in a protein called the CFTR (cystic fibrosis transmembrane conductance regulator) protein, which is a salt channel present in many organs of our bodies, including our sinuses, lungs, pancreas, gut, liver and reproductive organs. Salt goes through this channel, along with water, and this helps hydrate secretions and mucus. In CF, because of this dysfunctional salt channel (CFTR protein), thick mucus forms in the lungs, the pancreas and other organs, which then causes issues.



In the lungs, the mucus blocks the small airways, causing inflammation, recurrent infections and results in lung damage and difficulty breathing.

CF is usually diagnosed in childhood, but sometimes milder forms can be discovered in adulthood, especially when patients do not have frequent respiratory infections.

Sinus disease, pancreatitis, male infertility, diabetes and liver disease can also be symptoms associated with a late diagnosis of CF.



// What are the symptoms of CF?

UPPER AND LOWER RESPIRATORY TRACTS

Most individuals with CF will have had past upper and lower respiratory tract infections, ranging from sinusitis, bronchitis or pneumonia. About 10% of people with CF will have asthma, too.

Some will suffer from a lung collapse, or pneumothorax, but this is rarely one of the first symptoms that occurs.

The most common symptoms of CF are:

- Long-lasting cough (i.e., dry or wet cough)
- Frequent chest infections
- Wheezing
- Shortness of breath
- Frequent sinus infections

GASTROINTESTINAL TRACT

The pancreas plays a key role in helping digest fats and proteins, thanks to the enzymes it produces. It also secretes insulin, which is important in keeping blood sugar levels normal. In CF, the pancreas can get damaged and blocked by thick mucus.

When the pancreas is damaged by CF, it can cause pain attacks called pancreatitis.

When enough of the pancreas is damaged by CF, the body cannot properly absorb fat and protein contained in the diet. You can then have weight loss, fat malabsorption, large and oily bulky stool and become malnourished. This is called pancreatic insufficiency (exocrine function of the pancreas).

When a big enough portion of the pancreas is destroyed by CF, it no longer produces enough insulin, which results in high blood sugar. This is called CF-related diabetes, and the symptoms associated with high blood sugars are fatigue, weight loss, thirst, and frequent urination.

In CF, due to thick mucus in the gut and trouble absorbing fats, people can develop constipation and their intestine can become blocked transiently.

The most common gastrointestinal symptoms associated with CF are:

- Frequent, large, greasy and foul-smelling bowel movements
- Inability to gain weight despite being hungry all of the time and eating enough
- Poor growth
- Constipation and blocked intestine
- Recurrent inflammation of the pancreas (pancreatitis)
- Symptoms of high blood sugar such as being thirsty and frequent urination

REPRODUCTIVE SYSTEMS

Most CF patients with male sex organs are infertile. This is due to the absence of the vas deferens, which are the tubes that transport the sperm from the testicles to the ejaculatory ducts. This tube gets blocked by thick mucus before birth and does not form. However, there is still sperm production, and so these patients with CF can have biological children through procedures of sperm extraction and in vitro fertilization.

CF patients with female sex organs are not infertile, but they can sometimes have a harder time getting pregnant. This is most often correlated with how severe their lung disease is, and their nutritional status.

// How is CF diagnosed?

Most people with CF are diagnosed before the age of 2, and since 2018, newborns are screened for the condition in Québec. This means that many babies are diagnosed before they have symptoms, so they can begin receiving care as early as possible.

a. Basic tests and medical history



In adulthood, the diagnosis might be less obvious since a lot of the symptoms are milder and different than how CF presents in childhood. A good medical history, physical exam, basic blood tests and breathing tests will be helpful.

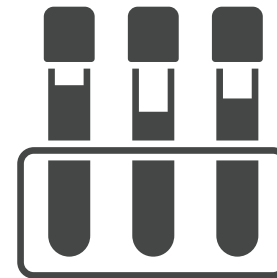
Other tests may include a CT scan of the chest or the sinuses and a sweat chloride test.

b. Sweat chloride test

The sweat chloride test is the “gold standard” test, meaning the best available test, to assess the function of the CFTR protein, the salt channel affected by CF.

During a sweat chloride test, sweat is taken from a small area of skin over the inner forearm and the amount of salt in that sweat is measured. Because of the abnormal function of the CFTR channel protein, people with CF have a lot of salt in their sweat: the dysfunctional CFTR protein does not reabsorb the salt in the skin properly, causing high levels of salt in the sweat.

c. Blood test



After the sweat chloride test, doctors will have you pass a genetic test to try to identify 2 mutations associated with the disease to confirm the diagnosis. More than 2,000 gene mutations have been found in CF. Most are quite rare, but a few are common, like the deltaF508 mutation that is found in at least 1 gene in 90% of individuals with CF.

These genetic tests are done from a regular blood test. We do the gene sequencing on the blood here at the MUHC but sometimes, we need to send another blood sample to the Canadian Referral Center for CFTR testing at the Sick Kids Hospital in Toronto for more advanced testing.

// Genetic counselling

Since CF is an autosomal recessive inherited disorder, a person needs to have 2 mutations in the affected gene (CFTR gene) to have the disease: 1 mutation from each parent.

For couples who want to have children, genetic testing is also important as more than 1 million Canadians are carriers of a CF gene: they have 1 mutation or mistake in the CF gene and have no symptoms. For every pregnancy when both parents are carriers, there is 1 chance in 4 that the child will have CF.

// Related diseases

In rare cases, we are not able to confirm whether someone has CF. Other medical conditions fall in the range of CF. When the sweat chloride test is positive, but the symptoms are not fully like those of CF, we might suspect a condition called CFTR-related disorder, where there is a dysfunction of the CFTR channel protein. Especially when only 1 organ is affected (pancreatitis, male infertility, isolated bronchiectasis), with no other CF symptoms.

// Living with CF

HOW IS CF FOLLOWED AND TREATED?

Treating chest infections is very important to prevent or slow down the long-term lung damage from CF.

a. Chest physical therapy

We will often ask our patients to participate actively in their treatment by doing airway clearance therapy (i.e., exercises that help clear the airways) or chest physiotherapy. This can be done in various ways, with the use of devices (i.e., TheraPEP™, Aerobika™, Flutter, Acapella™), manual therapy (clapping) and breathing exercises.

b. Inhaled medications

- Help the mucus to clear (i.e., hypertonic saline water)
- Thin the mucus (i.e., Pulmozyme™, a medication that contains enzymes that break down white blood cells in the mucus to make it less thick and easier to cough up)
- Reduce the amount of bacteria in the lungs (i.e., inhaled antibiotics, such as Tobramycin, Tobi™, Cayston™, Quinsair™, Colimycin)

c. Enzymes

Medications that help replace enzymes in the pancreas are also very important. Enzymes help the body absorb food and necessary nutrients. Enzymes have to be taken before every meal and snack. People with CF also have to take certain vitamins such as Vitamin A, D, E and K and minerals like calcium to ensure bone health. Maintaining normal weight is very important, as it has been shown to be linked to lung health. A big emphasis is put on this in our clinic

d. Antibiotics

Occasionally, intravenous antibiotics are required to treat acute infections. This sometimes requires a short hospital stay depending on the severity of the illness

e. CFTR modulators

Since 2015, new medications specifically designed to help with the underlying CFTR dysfunction are available. They are called CFTR modulators and will be acting on specific CFTR mutations. When eligible, certain patients may benefit greatly from such therapies. Currently, 4 medications have been approved in Canada: Kalydeko™, Orkambi™, Symdeko™ and Trikafta™.

DOCTOR VISITS

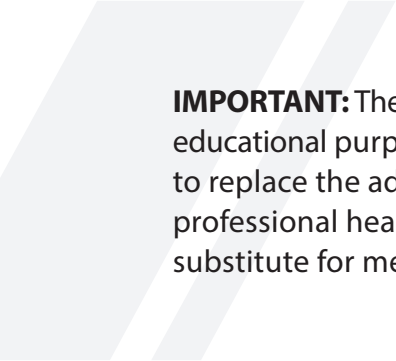
Depending on how severe the disease is, you should expect to see your lung doctor every 3 to 6 months and more often when there is an active issue. Most patients will also need to be followed by an Ear Nose and Throat doctor for their sinus disease and an endocrinologist if they suffer from CF-related diabetes. A nurse clinician, social worker, nutritionist and physiotherapist are also part of the multidisciplinary team to ensure your needs are met.

A visit to the lung doctor usually consists of a breathing test, a chest X-ray, a sputum collection for culture, and once a year, you should undergo a complete set of blood tests for a complete blood count, liver function, renal function, screening for diabetes, vitamin level, thyroid function, etc. Once every 2 to 5 years, you will also undergo a bone density to look for osteoporosis and an abdominal ultrasound to look at your liver.

// *What to expect?*

Once a diagnosis of cystic fibrosis is confirmed, you will meet with the care team. We will explain to you what CF is and map out your CF care strategy and discuss any of your concerns with you.

Being diagnosed with CF later in life is usually a sign of a much milder disease and you probably do not fit in the usual statistics associated with CF, so it is important to put that into perspective when reading about your disease.



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