

## Clinical update: Cystic fibrosis

### Overview

Cystic fibrosis (CF) is a genetic disorder affecting mostly the lungs but also the pancreas, liver, kidneys and intestine. It can also be known as mucoviscidosis (“Cystic fibrosis”, 2015) and two thirds of patients are diagnosed by one year of age. CF is a disease with a wide diversity of clinical presentation, severity of symptoms, and rate of disease progression in the organs involved and the clinical manifestations will vary with the patient's age at presentation (Sharma, 2014).

### Causes

Cystic Fibrosis (CF) is a recessive genetic condition resulting from inheriting two copies of faulty gene from both parents (Cystic Fibrosis Australia, n.d.). The faulty gene is called the cystic fibrosis transmembrane conductance regulatory gene (known as the CFTR gene) and is responsible for creating a protein that moves salt and water out of a cell. If the CFTR gene is defective, this results in a build-up of thick, sticky mucus in the body's tubes and passageways. These blockages damage the lungs, digestive system and other organs, resulting in inflammation (swelling) and repeated infections (NHS Choices, 2014). In the lungs, the mucus clogs the tiny air passages and traps bacteria, resulting in repeated infections which can cause irreversible lung damage and death. In the pancreas, the thick mucus also stops digestive enzymes from reaching the small intestine, which leads to difficulty with digesting fats and absorbing some nutrients. This means that people with CF can have problems with nutrition (Better Health Channel 2012).

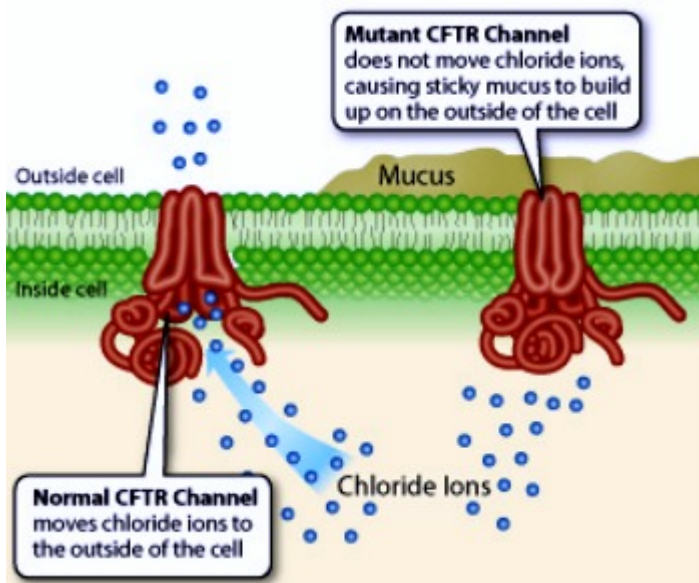


Figure 1: Cystic fibrosis – difference in the CFTR gene (University of Utah Health Sciences, n.d.)

### Prevalence

- One in every 2,500 Australian babies, male or female, of Northern European ancestry.
- About one in 25 Australians of Northern European ancestry are carriers for the CFTR gene mutation.
- CF is less frequent in Southern European and Middle Eastern populations, and is rare or absent in Asian populations (Genetics in Family Medicine, 2007).

### Complications

CF has a wide variety of complications across multiple body systems. These are related to the mucus deposits in various organs, leading to infections and difficulty with absorption of nutrients and vitamins.

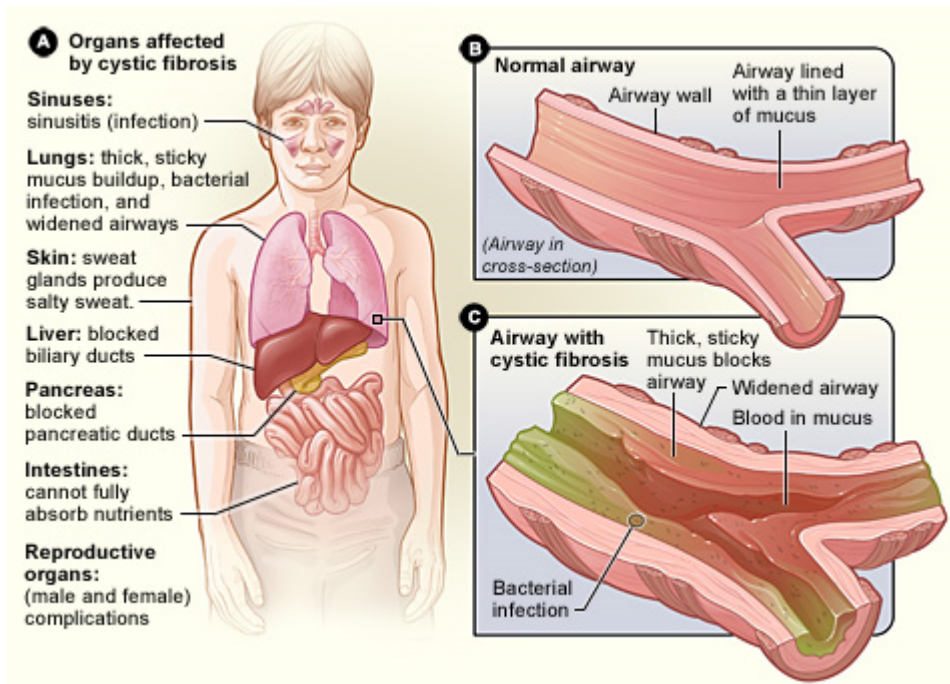


Figure 2: What are the signs and symptoms of Cystic fibrosis? (National Heart, Lung, and Blood Institute, 2013)

## Manifestations

The manifestations of CF affect multiple body systems including:

- respiratory
- gastrointestinal
- genitourinary
- endocrine.

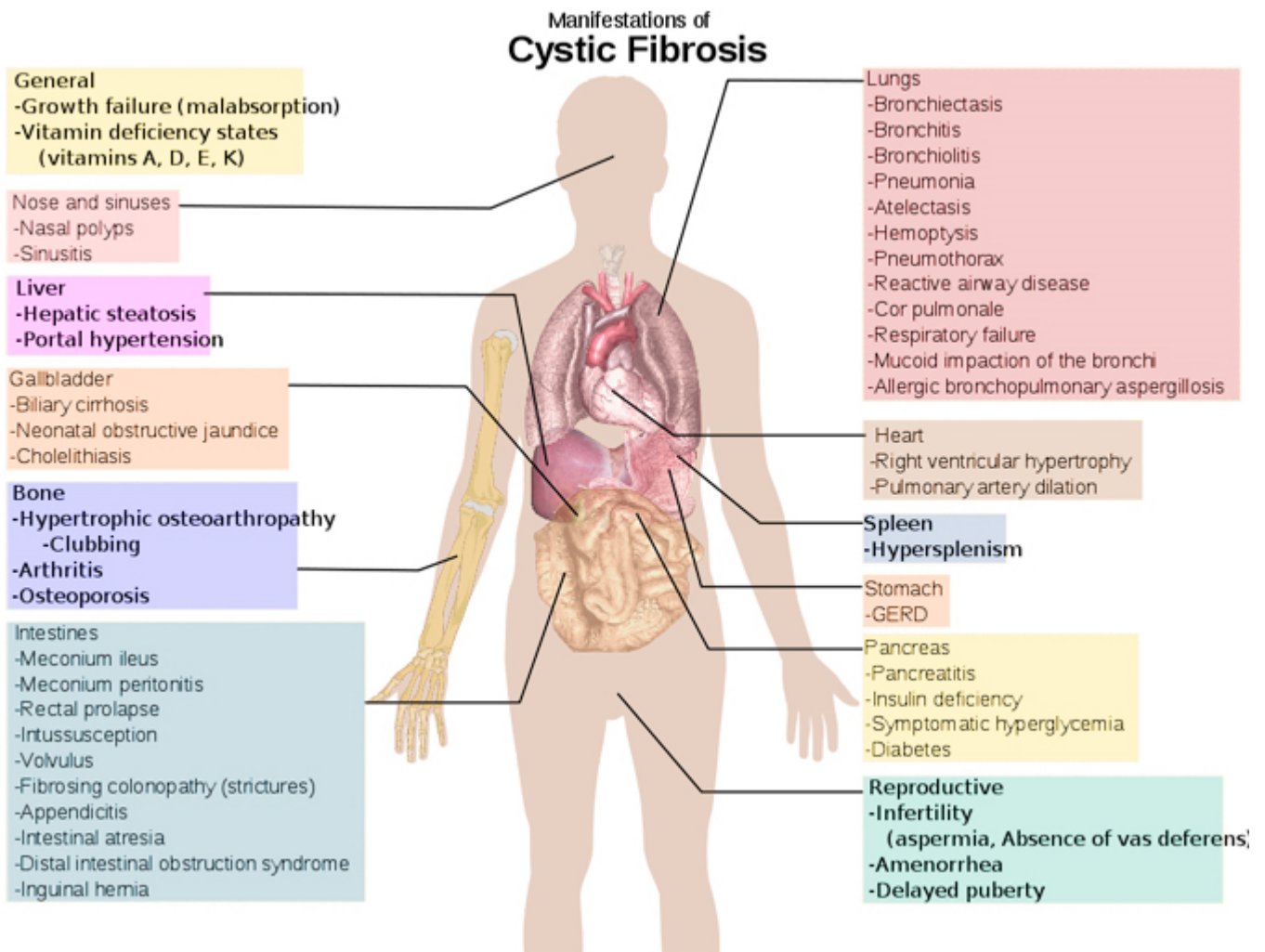


Figure 3: Manifestations of cystic fibrosis (Newson, 2012)

The image above identifies the potential manifestations of CF. Additional manifestations which may occur include:

- pulmonary hypertension
- fatty liver
- liver failure
- cholecystitis
- rickets (Sharma, 2014).

### Management

CF has no cure, however, treatments have greatly improved in recent years. The goals of CF treatment include:

- preventing and controlling lung infections
- loosening and removing thick, sticky mucus from the lungs
- preventing or treating blockages in the intestines
- providing enough nutrition preventing dehydration (a lack of fluid in the body) (National Heart, Lung, and Blood Institute, 2013).

Treatment options include:

- antibiotics – to treat chest and lung infections
- intensive physiotherapy – to clear mucus from the lungs
- enzyme replacement capsules with food to aid digestion
- salt vitamin supplements

- bronchodilators – a type of medication that helps expand the airways inside the lungs, making it easier to breathe
- inhaled mucus-clearing treatments – such as Pulmozyme, hypertonic saline (a strong salt solution) and mannitol dry powder
- exercise – to help clear the airways and build core strength (Cystic Fibrosis Australia, n.d.; NHS Choices, 2014).

In some cases a lung transplant may eventually be required if the lungs become extensively damaged.

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