

Disease A - Cystic Fibrosis

Cause or main causes

Cystic Fibrosis is caused by a change, or mutation, in a gene called CFTR (Cystic Fibrosis Transmembrane conductance Regulator). This gene controls flow of salt and fluids in and out of the cells in your body. If the gene doesn't work the way it should a sticky mucus builds up in your body.

To get Cystic Fibrosis, you have to have inherited the mutated copy of the gene from both parents. 90% of people affected have at least one copy of the F508del mutation.

If you inherit only one copy, you won't have any of the symptoms, but you will be a carrier of the gene, meaning there is a chance you could pass it to your children.

Symptoms and diagnosis

- Trouble with bowel movements or frequent, greasy stools
- Wheezing or trouble breathing
- Frequent lung infections
- Infertility, especially in men
- Trouble growing or gaining weight
- Skin that tastes very salty

Early diagnosis means early treatment and better health later in life.

Blood test - This checks the levels of immunoreactive trypsinogen (IRT). People with Cystic Fibrosis have higher levels of it in their blood.

DNA test - This looks for mutations to the CFTR gene.

Sweat test - It measures the salt in your sweat. Higher than normal results suggests Cystic Fibrosis

Some people who weren't tested as newborns aren't diagnosed until they become adults.

Current treatments - how they work and what they do

There is no cure for Cystic Fibrosis, but there are medications and other therapies that can ease symptoms.

Medications

- **Antibiotics** can prevent or treat lung infections and help the lungs to work better. You can get them as pills, in an inhaler, in a shot.
- **Anti-inflammatory medicines** includes ibuprofen and corticosteroids.
- **Bronchodilators** comes from an inhaler, they relax and open the airways.
- **Mucus thinners** helps get gunk out of the airways, comes from an inhaler.
- **CFTR modulators** helps the CFTR work like it should, makes your lungs work better and also helps you gain weight
- **Combination therapy** new medication elexacaftor/ivacaftor/tezacaftor (Trikafta). Combines three CFTR modulators to target the CFTR protein and make it work precisely.

Airway clearance techniques

- **Chest therapy or percussion** involves tapping or clapping on the chest or back to clear mucus from the lungs. Someone else does this for you.
- **Oscillating devices** breathe into a special device that oscillates, or vibrates, the airways. Loosens mucus and makes it easier to cough up.

Physical therapy for CF

- **Autogenic drainage** this is done by breathing out hard or huffing. This moves mucus from the smaller airways to the central airways making it easier to get out.
- **Active cycle of breathing** this controls the breathing and relaxes the upper chest and shoulders, which can help clear the mucus and prevent airway blockages. To do this you breathe in deeply, hold it, and then you huff for different lengths of time

Prevention

As Cystic Fibrosis is a genetic disease there is no prevention for the disease.

Disease B - Tuberculosis

Cause or main causes

Tuberculosis is caused by a type of bacterium called Mycobacterium Tuberculosis

Symptoms and diagnosis

General symptoms of TB

- Lack of appetite and weight loss
- A high temperature
- Night sweats
- Extreme tiredness and Fatigue

TB that effects the lung

- A persistent cough that lasts more than 3 week and typically brings up phlegm, which may be bloody
- Breathlessness that gets worse as time goes on

Diagnosis

It is difficult to diagnose and several tests may need to be done. You may have a chest x ray to look for changes in the lungs that might suggest TB. Samples of phlegm will also often be taken to be checked for the presence of TB bacteria.

Current treatments - how they work and what they do

You will be prescribed with at least a 6 month course of a combination of antibiotics if you've been diagnosed with pulmonary TB.

The usual treatment is

2 antibiotics (isoniazid and rifampicin) for 6 months

2 additional antibiotics (pyrazinamide and ethambutol) for the first 2 months of the 6-month treatment period

It might be several weeks before you start to feel better, The exact length of time depends on your overall health and the severity of the Tuberculosis. After 2 weeks of taking the antibiotics most people begin to feel better and become no longer infectious, but it is important to keep going on with the prescribed antibiotics and complete taking them as the instructions say.

Taking the medication for 6 months is the best way to ensure all of the TB bacteria is killed.

Prevention

There is the vaccine called Bacillus Calmette-Guerin (BCG). It is one of the most widely used of all current vaccines, and it reaches more than 80% of all new born children and infants in countries where it is part of the national childhood immunisation programme. The BCG vaccine is more generally used to protect children than to interrupt transmission between adults.