Among people said to be healthy the AAT serum level measures between 1.5 and 3.5 g/L. For people with a serum level measuring below 1.2 g/L, genetic tests should be initiated to identify the presence or absence of genetic variants associated with AAT deficiency.

In Quebec, AAT serum levels can be performed in most hospitals. A comprehensive program designed to screen for AAT deficiency (serum levels and/or genetic tests) is available at the Centre Hospitalier Universitaire de Montréal (CHUM) as well as the McGill University Health Centre (MUHC).

TREATMENTS AND PREVENTION

There is presently no known cure for this disease.

However, people with this disease can slow down its progression by adjusting their lifestyles and opting for treatments that will help alleviate the symptoms :

- Quit smoking ;
- Avoid professional activities exposing you to environmental pollutants like: sulfur dioxide, nitrogen oxide and airborne dust particles ;
- Undertake a pulmonary rehabilitation program ;
- Undergo vaccination ;
- Use bronchodilators, corticosteroid inhalers, use oxygen or other medicated treatments of this type as recommended by your doctor.

AUGMENTATION THERAPY

This therapy makes it possible to raise AAT levels in the blood. It's a weekly injection that the Canadian Thoracic Society (CTS) recommends, provided that certain criteria are met. It involves administering an infusion of purified human AAT as a means of raising AAT levels in the blood of people with an AAT deficiency associated with emphysema. The main purpose of this therapy is to slow down the progression of this pulmonary disease.

According to CTS guidelines, the following people are eligible for this type of therapy. The eligibility criteria are as follows :

- Non-smoker ;
- AAT blood level less than 0.6 g/L and/or linked to a congenital AAT deficiency ;
- Diminished pulmonary capacity (25-80% FEV1 spirometry)

Intravenous administration of an AAT supplement is useless in patients with a liver disease caused by an AAT deficiency. With regard to the administration of Prolastin-C, a protein derived from highly purified plasma product, it is available to people with a congenital AAT deficiency, clearly suffering from clinically-recognizable emphysema.

N.B. : In the case of companies that split proteins derived from human plasma for therapeutic purposes, regulatory authorities, like Health Canada apply very strict standards, which makes the use of these products not only free of risk, but also very safe where the potential transfer of diseases is concerned.

THE QUEBEC LUNG ASSOCIATION

The Quebec Lung Association was incorporated in 1938 and is the only non-profit organization promoting respiratory health. Its mission is to fight lung disease through education, prevention, rehabilitation, support for the people affected and their families, as well as research on respiratory diseases.

The QLA also addresses environmental factors that pose risks to respiratory health.

Through its work to improve the respiratory health of Quebecers and the living conditions of people suffering from respiratory illnesses, and by encouraging them to take charge of their health, the Quebec Lung Association acts directly on the condition of people of all ages, which has a direct impact on public health.

Contact us by calling
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ALPHA-1 ANTITRYPSIN DEFICIENCY

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ALPHA-1 ANTITRYPSIN DEFICIENCY

Alpha-1 antitrypsin (AAT) deficiency is one of the world's most common genetic anomalies.

Over 7,000 Canadians are estimated to have one of the severe forms of AAT deficiency, although fewer than 5% of them are diagnosed. This deficiency is then likely to significantly affect lung condition.

AAT is a protein mainly produced by the liver and it is present in practically the entire body. Its main function is to protect lung tissue by destroying certain substances released by white cells (immune system cells), including elastase.

In people with an AAT deficiency, elastase cannot be destroyed by AAT and thus can cause the loss of elasticity of the lungs.

In isolated cases, AAT deficiency can be linked to an accumulation of AAT in the liver, thus triggering the onset of various liver diseases.

However, the lungs and liver are practically never affected at the same time.

WHAT IS A GENETIC DISEASE ?

Also known as a hereditary disease, a genetic disease is the complex result of modifying one or several genes.

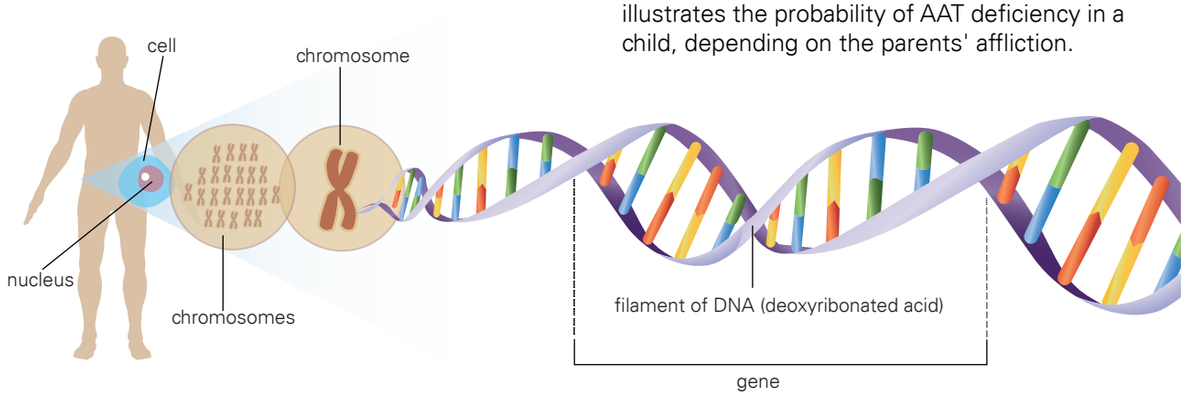
It renders some of the body's cells unable to correctly fulfill their role. Its particularity is that it is vertically transmitted, i.e. parents to children.

To date, over 6,000 different genetic diseases have been identified.

CAUSES

AAT deficiency is a hereditary disease. At least one of the two parents has to be a carrier or be afflicted with AAT deficiency in order to pass it on to his/her child. It is the only way someone can develop this disease. However there are aggravating factors, the main one being cigarette smoke, followed by environmental pollutants.

Over 200 different variants of the gene responsible for AAT deficiency have been identified to date. Some of them being linked to a severe deficiency that leads to the onset of emphysema. The following table clearly illustrates the probability of AAT deficiency in a child, depending on the parents' affliction.



	Possible parent combinations (father and mother)				
Child profile					
	50%	25%	---	---	---
	50%	50%	50%	100%	---
	---	25%	50%	---	100%

Legend : **Healthy** **Carrier** **Afflicted**

SIGNS AND SYMPTOMS

AAT deficiency can cause symptoms similar to those of chronic obstructive pulmonary disease (COPD) and especially emphysema.

As a result, many people fail to realize that they have an AAT deficiency until severe symptoms appear. The signs and symptoms include the following:

- Shortness of breath (dyspnea) ;
- Coughing ;
- Wheezing.

Respiratory symptoms may appear much earlier in patients with AAT deficiency who smoke.

Other symptoms of a non-respiratory origin may appear, such as jaundice of the eyes, ascites (swelling of the abdomen) and other unexplained liver problems.

DIAGNOSIS AND SCREENING

According to the guidelines of the Canadian Thoracic Society (CTS), a test designed to screen for AAT deficiency is recommended for :

- Any person with a COPD diagnosed before the age of 65 ;

or

Any person with a COPD, presenting a null-to-moderate history of smoking (having smoked approximately one pack of cigarettes a day for 20 years or less) ;

- Any person with a family history of AAT deficiency.

The first step in diagnosing the disease involves drawing a blood sample that will serve to measure AAT levels. This pooled test is performed using reference methods and algorithms that make it more reliable.