

What is Alpha-1 Antitrypsin (AAT)?

- ✓ Alpha-1 antitrypsin (AAT) is a protein manufactured primarily in the liver.
- ✓ AAT protects tissues throughout the body. This document focuses on how AAT protects the lungs.
- ✓ AAT protects the lungs from enzymes that can injure the lungs, such as neutrophil elastase.
- ✓ The amount of neutrophil elastase in the lungs is especially high when:
 - You have an infection in your lungs
 - You are exposed to smoke or other inhaled irritants such as dust or fumes
- ✓ Neutrophil elastase can be helpful. It breaks down bacteria and damaged tissue. However, neutrophil elastase also can cause injury to healthy lung tissue.
- ✓ Normally there is a balance between AAT and neutrophil elastase in the lungs. When in balance, neutrophil elastase breaks down bacteria and damaged tissue without injuring healthy lung tissue.

What is AAT Deficiency?

- ✓ Individuals with AAT deficiency have a reduced level of AAT circulating in their blood, or have AAT that is not functional.
- ✓ AAT deficiency is a hereditary condition. Everyone inherits two gene alleles for AAT: one from their mother and one from their father.
- ✓ A genetic variant that produces less AAT than normal, produces AAT that is not functional, or produces AAT that gets trapped in the liver is referred to as a deficiency variant (or deficiency allele).
- ✓ The extent to which you may have an increased risk for health problems depends on:
 - Whether you have 1 deficiency variant or 2 deficiency variants
 - The specific deficiency variant(s) you have inherited (for example, Z, S, F, or Null)

Why do Adults with AAT Deficiency Have Increased Risk of Lung Disease?

- ✓ The increased risk of lung disease among adults with AAT deficiency is due to a shortage of functional AAT circulating throughout the body, especially in the lungs.
- ✓ AAT protects the lungs by inactivating neutrophil elastase before it can injure healthy lung tissue.
 - A primary way that neutrophil elastase injures the lungs is by destroying elastin
 - Elastin gives tissues the ability to stretch and then return to their normal state
 - Lung tissue is especially rich in elastin
 - With decreased AAT protection, lung damage accumulates over time and progresses slowly
- ✓ Adults with AAT deficiency have an increased risk of:
 - Chronic obstructive pulmonary disease (COPD), which includes emphysema (destruction of the walls of the air sacs of the lung)
 - Bronchiectasis (enlarged bronchial tubes)
- ✓ These lung diseases are typically seen in adults with AAT deficiency, not children. However, exposures in childhood can increase the risk of developing lung disease as an adult.

Additional Information

- ✓ AlphaNet's Big Fat Reference Guide (BFRG) has several chapters about lung health. Anyone can access [AlphaNet's BFRG](#). AlphaNet Subscribers can access the BFRG through their [Subscriber Portal](#).
 - Chapter 1 (Alpha-101) has information about how AAT deficiency increases risk of disease
 - Chapter 10 (Staying Healthy with Lung Disease) has steps you can take to protect your lungs and slow the progression of lung disease
- ✓ A video that describes AAT deficiency and explains how it increases the risk of developing disease is available on the Alpha-1 Foundation website at <https://vimeo.com/247506044>.
- ✓ A pair of gene alleles is called a genotype. Lung disease risk differs by genotype. You can enter your genotype in the [Alpha-1 Alleles website](#) and learn about your lung disease risk.
- ✓ AlphaNet's Skinny Little Reference Guide [Staying Healthy: Management of Environmental Risk Factors](#) can help you protect your lungs by reducing your exposure to risk factors for lung disease.