

What Is a “Deficiency Variant”?

- ✓ Everyone inherits one alpha-1 antitrypsin (AAT) gene from their mother and one AAT gene from their father. Variants of each gene can have no effect or can lead to deficient/dysfunctional AAT.
 - ❑ The normal gene is called “M”.
 - ❑ There are many deficiency variants. The most commonly-identified deficiency variants are called “Z” and “S”.
- ✓ A deficiency variant produces less AAT than a normal gene and/or produces AAT that gets trapped in the liver and cannot travel through the body to protect the lungs.
- ✓ Individuals with deficiency variants may have a higher risk of developing lung and liver disease.
- ✓ Information about the health risks associated with different variants (occurring at specific places in the gene, called “alleles”) is available at the Alpha-1 Alleles website found [here](#).

Should My Family Members Consider Getting Tested?

- ✓ If you have at least one deficiency variant, family members should consider getting tested because:
 - ❑ They may have a higher risk of developing lung and liver disease. The risk of developing medical problems can be reduced through health behaviors such as avoiding smoking.
 - ❑ They can pass a deficiency variant to their children.
- ✓ Which family members should consider getting tested?
 - ❑ The highest priority is parents, siblings, and children.
 - ❑ Then, consider testing extended family members such as aunts, uncles, and cousins.
- ✓ Additional information about testing recommendations is available [here](#).

How Can Family Members Get Tested?

- ✓ Testing uses a small sample of blood or a small sample of cells that are collected with a cheek swab.
- ✓ Individuals can request a free, confidential test kit. Results are returned to the individual who requested the test kit. One option is the Alpha-1 Coded Testing (ACT) study through the Alpha-1 Foundation. Information is available [here](#) or by calling 1-855-476-1227.
- ✓ Individuals can work with their healthcare provider to get tested. If testing is ordered by a healthcare provider, results are returned to the healthcare provider.
- ✓ AlphaDetect is a non-profit subsidiary of the Alpha-1 Foundation that was founded in 2025. AlphaDetect plans to provide free genetic testing starting in mid-2026. Information is available [here](#).

What are Possible Risks of Testing?

- ✓ There are some potential risks of testing. These risks include:
 - ❑ It can be stressful to learn that you or a family member have a genetic condition.
 - ❑ Genetic discrimination is possible. However, protections are in place through the Genetic Information Nondiscrimination Act (GINA). More information about GINA is available [here](#).
- ✓ Individuals may want to talk with a genetic counselor before and/or after getting tested.
 - ❑ A genetic counselor can explain the potential risks and benefits of testing.
 - ❑ The Alpha-1 Foundation provides free genetic counseling services. The phone number for these services is 1-855-476-1227.

What are Possible Benefits of Testing?

- ✓ Test results can help individuals make decisions that protect their health, which include:
 - ❑ Making lifestyle changes such as quitting smoking and avoiding excessive alcohol use.
 - ❑ Participating in education and research through the Alpha-1 Foundation, the Alpha-1 Research Registry, or other organizations.
 - ❑ Learning about additional treatment options that may be available to treat lung disease.
 - ❑ Making more informed decisions about having children based on the likelihood that children may inherit deficiency variants.