AlphaID™ At Home
Genetic Health Risk Service Report

Hi Jane,

You have taken an active step for your health and your family's health. This is your **AlphaID™ At Home Genetic Health Risk Service Report.** It contains your genetic results.

What is alpha-1 antitrypsin deficiency (AATD)?

AATD is a genetic condition caused by genetic variants in a gene called *SERPINA1*. It is passed down from parents to their children. Most commonly, AATD leads to lung and/or liver disease.

For more information about AATD, how common it is, its symptoms, and treatment, see **pages 7 and 8**.

What is the Service for?

DETECTS 14 variants in the *SERPINA1* gene linked to AATD. These 14 variants explain 95% of AATD cases.

DETERMINES if your genetics increases your risk of developing lung and/or liver disease linked to AATD compared to the general population.

This report will help you understand:

- Your genetic result (either 0, 1, or 2 variants detected in the *SERPINA1* gene)
- Your risk of developing lung and/or liver disease linked to AATD compared to the general population

Limitations of the Service

- **Does NOT detect all possible variants** linked to AATD
- **It is NOT a substitute for an appointment** with a healthcare professional
- **Does NOT diagnose any disease or condition.** Only a healthcare professional can diagnose a disease or condition
- **Does NOT determine if you have or will develop lung and/or liver disease** linked to AATD during your lifetime

Jane, this is your genetic result for AATD

You have

2 Variants:

PI*Z and PI*Z
in the *SERPINA1* gene
linked to AATD

Lung disease risk

- You are at ***increased risk*** of developing lung disease linked to AATD compared to the general population*

Liver disease risk

- You are at ***slightly increased risk*** of developing liver disease linked to AATD compared to the general population*

*General population is defined as all adults who live in the United States.

Please see Glossary of Terms on page 24 for a definition of **Genetic result**.

Important Next Step

Share your results report with a healthcare professional.

Healthcare professionals can answer questions you may have about your results, risk, and how they may apply to your health. They should know you were tested for AATD.

For more information about next steps, see page 5.

For information about more resources, see page 14.

Additional Non-Genetic Risk Factors

There are other non-genetic risk factors which may increase your risk of developing lung and/or liver disease linked to AATD. These factors can include lifestyle choices, work/home environment, and your exposure to infections. Changing behavior to avoid these factors may reduce your risk level.

Smoking can increase your risk of developing lung disease

Alcohol consumption can increase your risk of developing liver disease

Environmental pollution at work, home, and around you can increase your risk of developing lung disease

Obesity can increase your risk of developing liver disease

Liver and lung infections can increase your risk of developing liver and lung disease

A personal or family history of lung and/or liver disease can increase your risk of those diseases. This risk factor cannot be changed.

About Your Results and Your Family

You share genetics with your biological family members. So, your result may be important for their health, too.

You have **2 variants, PI*Z and PI*Z,** in the *SERPINA1* gene linked to AATD, so:

- You inherited 1 variant from each of your parents
- You will pass 1 of your 2 variants to your children
- If your child's other biological parent has a *SERPINA1* variant, your child could inherit two variants, one from each parent. This may put them at risk of developing lung and/or liver disease linked to AATD.

Ethnicity and AATD

AATD occurs in people of all ethnicities worldwide. However, it is most common in people of European descent, because they are more likely to have variants in the *SERPINA1* gene. AATD affects about 1 in 1,500 to 3,500 people of European descent.

People of non-European descent can also have variants in the *SERPINA1* gene that increase their risk of developing lung and/or liver disease linked to AATD. This means even if you are not a European descent, your genetic risk report still applies to you.

Important Next Steps

1. **Share your results report with a healthcare professional.**
   Healthcare professionals can answer questions you may have about your results, risk, and how they may apply to your health. They should know you were tested for AATD. You should also inform a healthcare professional if you:
   - Have symptoms of lung or liver disease (see page 8 for more information)
   - Have a personal or family history of lung or liver disease
   - Are feeling anxious, uncertain, or concerned about your genetic result or risk
   - Have questions about any risk factors

2. **Consider sharing this report with your family members.**
   Since you share genetics with biological family members, your result may be important for their health as well. Talk to a healthcare professional about family testing.

3. **Use the links below to follow through on these next steps:**
   - To find a doctor with experience in AATD from the Alpha-1 Foundation's Clinical Resource Centers, visit **www.alpha1.org/alphas-friends-family/resources/find-an-alpha-1-specialist**
   - To find a healthcare professional near you with experience testing for AATD, please visit **www.AlphaFindADoctor.com**
   - To find a genetic counselor, visit **https://findageneticcounselor.nsgc.org/**

For information about more resources, see page 14.

AlphaID™ At Home
Genetic Health Risk Service Report
Additional Information

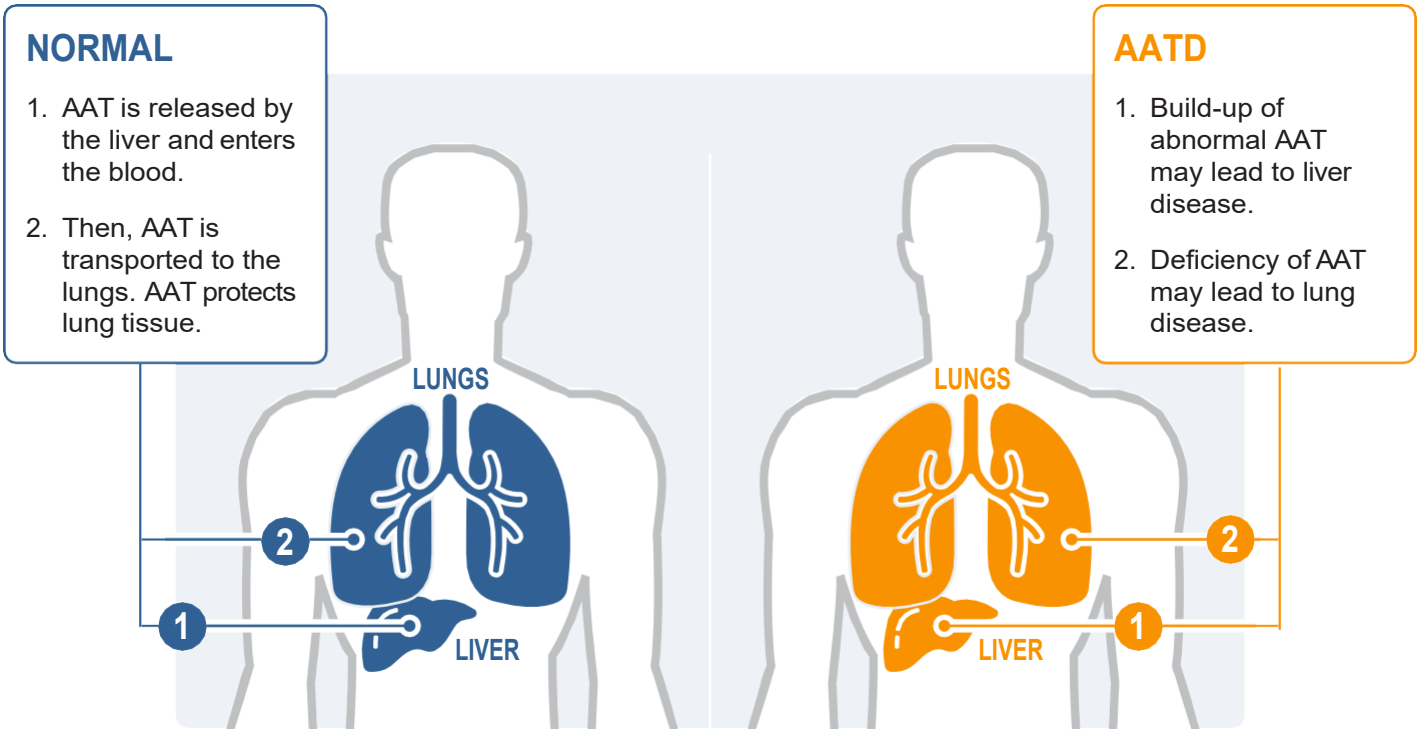
Scientific details of alpha-1 antitrypsin deficiency (AATD)[1,2]

Alpha-1 antitrypsin deficiency is a genetic condition. It is passed from parents to their children. AATD may lead to various health conditions. Most commonly, AATD leads to lung and/or liver disease.

AATD is caused by certain genetic variants in the *SERPINA1* gene. These variants cause low levels of a protein called **alpha-1 antitrypsin (AAT)**. Low levels of the AAT protein lead to AATD.

How AATD Impacts the Body[1]

The *SERPINA1* gene instructs the liver to make AAT. Then, it enters the blood and goes to the lungs. AAT plays a critical role in safeguarding the lungs. Low levels of AAT may cause lung disease linked to AATD. This condition can also lead to a build-up of AAT in the liver. This build-up may cause liver disease linked to AATD.



Adapted image from the following source: http://www.beaumont.ie/index.jsp?p=103&n=142&a=356

Signs and Symptoms Linked to AATD[3,4]

- Shortness of breath and wheezing
- Chronic cough
- Lung disease, including chronic obstructive pulmonary disease (COPD), emphysema, chronic bronchitis, and bronchiectasis
- Liver disease, including cirrhosis, jaundice, hepatic enzyme elevations, chronic hepatitis, and liver scarring (fibrosis)

Genetic Health Risk of AATD

- 2% to 3% of patients with COPD in the United States are estimated to have AATD[1]
- More than 90% of the estimated 100,000 people in the US with AATD don't know they have it[5,6]
- The percentage of patients with liver disease who have AATD is not known

Guidelines for AATD

The COPD Foundation, the World Health Organization, the American Thoracic Society, and the GOLD COPD guidelines recommend testing all COPD patients for variants linked to AATD.[1,7,8,9] A second method to confirm an AATD diagnosis should be considered.[8]

Testing also extends to people with unexplained liver disease.[1,8]

Treatment for AATD

It is important to know that while there is no cure for AATD, treatment options may be available. Augmentation therapy is a treatment option for lung disease linked to severe AATD. It raises the level of AAT protein in the blood. Also, it is important that people with AATD avoid smoking.[1,7,8,9]

There are no current specific treatment options for liver disease linked to AATD. The treatment options are the same ones that are used for liver diseases in general. People with liver disease linked to AATD should avoid alcohol consumption.[1,2,10]

What the Service Is For

The intended purpose of the AlphaID™ At Home Genetic Health Risk Service:

- **Detects** 14 variants in the *SERPINA1* gene linked to AATD. These 14 variants explain 95% of AATD cases[11]
- **Determines** if your genetics increases your risk of developing lung and/or liver disease linked to AATD compared to the general population

Read information on the **14 genetic variants**

Read information on the **risk categories** used by the Service

Read the **Package Insert** for more information about special considerations for testing, and clinical and analytical performance of the Service

Your Lung and Liver Disease Risk

Lung disease

- You are at ***increased risk*** of developing lung disease linked to AATD compared to the general population

  Your chance of developing lung disease linked to AATD is higher than that of the general population. This does not mean that you will develop lung disease.

Liver disease

- You are at ***slightly increased risk*** of developing liver disease linked to AATD compared to the general population

  Your chance of developing liver disease linked to AATD is slightly higher than that of the general population. This does not mean that you will develop liver disease.

Read information on the **risk categories** used by the Service

Important Next Step

Share your results report with a healthcare professional.

Healthcare professionals can answer questions you may have about your results, risk, and how they may apply to your health. They should know you were tested for AATD.

For more information about next steps, see **page 13**.

For information about more resources, see **page 14**.

Additional Non-Genetic Risk Factors

There are other non-genetic risk factors which may increase your risk of developing lung and/or liver disease linked to AATD. These factors can include lifestyle choices, work/home environment, and your exposure to infections. Changing behavior to avoid these factors may reduce your risk.

Smoking[1,2]

Cigarette smoke contains certain chemicals. In people with AATD, these chemicals can lead to inflammation of the lungs. Studies show that smokers with AATD have lung symptoms earlier than nonsmokers. They also have a faster decline in lung function. Severe symptoms are seen most in current or former cigarette smokers. Some smokers and many nonsmokers may not show symptoms and still have AATD.

Environmental pollution[1,51]

Research suggests that exposure to pollution, gas, fumes, and dust where you work or live is a risk factor. It may affect lung function.

More studies are needed to understand the effects of environmental exposure on lung disease linked to AATD.

Alcohol consumption[1,2,10]

Drinking alcohol can worsen liver disease. So, alcohol consumption should be avoided in people with AATD and liver disease. This may be a risk factor for liver disease linked to AATD, although the evidence is still unclear. More studies are necessary to establish the role of alcohol consumption and its association with liver disease linked to AATD.

Obesity[52]

Obesity is a lifestyle risk factor for liver disease. Obesity may cause diabetes, high blood pressure, and increased cholesterol. This can result in a fatty liver. Fatty livers are not good for those with AATD.

Lung and liver infections[1,53]

Respiratory infections can damage the lungs. Controlling these infections may benefit people with AATD. Hepatitis infections damage the liver. This may worsen liver disease caused by AATD. A healthcare professional may recommend preventive strategies for people with AATD. This may include vaccinations for influenza, pneumococcus, and hepatitis A and B.

Personal or family history of lung and/or liver disease[1]

A personal or family history of lung and/or liver disease can increase your risk of these diseases. This risk factor cannot be changed.

A personal history of lung symptoms such as shortness of breath and wheezing may increase the risk of developing lung disease in people with AATD. The risk of developing lung and liver disease can also depend on family history. If these diseases run in the family, this may increase the chances of similar diseases in other family members.

What Your Genetic Results Can Mean for Your Family

AATD is a genetic condition. It is passed from your biological parents to you. You can pass AATD on to your children. Everyone has 2 copies of the ***SERPINA1*** gene. One copy of this gene was passed to you from each of your parents. Since you share genetics with your family, your result may be important for their health, too.

You have **2 variants** in the ***SERPINA1*** gene linked to AATD. This means:

- You inherited 1 variant from each of your parents
- You will pass 1 of your 2 variants to your children
- If your child's other biological parent has a *SERPINA1 variant,* your child could inherit two variants, one from each parent. This may put them at risk of developing lung and/or liver disease linked to AATD.

The American Thoracic Society and the COPD Foundation recommend testing for variants linked to AATD in family members—parents, siblings, and children—of a person with AATD.[1,8]

Limitations of the Service

- It is not a substitute for an appointment with a healthcare professional. We strongly recommend you consult with a healthcare professional if you have any questions or concerns about your result or health
- It does not diagnose any disease or condition. Only a healthcare professional can diagnose a disease or condition
- It does not determine if you have or will develop lung and/or liver disease linked to AATD during your lifetime
- It cannot be used to make healthcare decisions. It does not tell you anything about your overall health. Only a healthcare professional can help you with healthcare decisions
- It detects 14 variants in the *SERPINA1* gene linked to AATD. These 14 variants explain 95% of AATD cases. It does not detect all possible variants linked to AATD
- There may be other, non-genetic factors that affect your risk. The Service does not determine your overall risk of developing lung and/or liver disease
- Other companies that offer AATD testing may detect different or fewer variants linked to AATD. You may get different genetic results using a test from a different company
- The laboratory may be unable to process every person's sample. If this happens, you will receive a notice and another AlphaID™ At Home Saliva Collection Kit so that you can provide a new sample to the laboratory

Ethnicity and AATD[1,2]

AATD occurs in people of all ethnicities worldwide. However, it is most common in people of European descent, because they are more likely to have variants in the *SERPINA1* gene. AATD affects about 1 in 1,500 to 3,500 people of European descent.

People of non-European descent can also have variants in the *SERPINA1* gene that increase their risk of developing lung and/or liver disease linked to AATD. This means even if you are not of European descent, your genetic risk report still applies to you.

Important Next Steps

1. **Share your results report with a healthcare professional.**
   Healthcare professionals can answer questions you may have about your results, risk, and how they may apply to your health. They should know you were tested for AATD. You should also inform a healthcare professional if you:
   - Have symptoms of lung or liver disease (see page 8 for more information)
   - Have a personal or family history of lung or liver disease
   - Are feeling anxious, uncertain, or concerned about your genetic result or risk
   - Have questions about any risk factors

2. **Consider sharing this report with your family members.**
   Since you share genetics with biological family members, your result may be important for their health as well. Talk to a healthcare professional about family testing.

3. **Use the links below to follow through on these next steps:**
   - To find a doctor with experience in AATD from the Alpha-1 Foundation's Clinical Resource Centers, visit **http://www.alpha1.org/alphas-friends-family/resources/find-an-alpha-1-specialist**
   - To find a healthcare professional near you with experience testing for AATD, please visit **www.AlphaFindADoctor.com**
   - To find a genetic counselor, visit **https://findageneticcounselor.nsgc.org/**

For information about more resources, see page 14.

Additional Resources

Alpha-1 Foundation

The Alpha-1 Foundation is the only national organization dedicated to developing a cure for alpha-1 antitrypsin deficiency and improving the quality of life for patients and their families. Acting as a patient advocate, the foundation helps the AATD community better connect to one another – strengthening the support and care networks they need. **https://www.alpha1.org** | (877) 228-7321 ext. 321

AlphaNet

AlphaNet is a not-for-profit organization providing innovative health management services and customized care to the AATD community via care coordinators who are AATD patients themselves. **https://www.alphanet.org** | (800) 577-2638

COPD Foundation

The COPD Foundation is a not-for-profit organization established to speed innovations that will make treatments more effective and affordable, undertake initiatives that result in expanded services for COPD patients, and improve the lives of patients with COPD.
https://www.copdfoundation.org

American Liver Foundation

American Liver Foundation provides community education programs, funds research, and supports advocacy related to liver disease.
https://liverfoundation.org/

MedlinePlus

The National Institutes of Health provide consumer-friendly information about the effects of genetic variation on human health.
https://medlineplus.gov/genetics/condition/alpha-1-antitrypsin-deficiency

References

View the **References** for additional information

View the **Educational Module** for more information about genetics

Frequently Asked Questions

1. What is alpha-1 antitrypsin deficiency (AATD)?

AATD is a genetic condition. It is passed from parents to their children. AATD may lead to various health conditions. Most commonly, AATD leads to lung and/or liver disease.

AATD is caused by certain genetic variants in the *SERPINA1* gene. These variants cause low levels of a specific protein. This protein is called alpha-1 antitrypsin (AAT). AAT protein is released by the liver, enters the blood, and is transported to the lungs. AAT plays a critical role in safeguarding the lungs. Low levels of AAT may cause lung disease linked to AATD. This condition can also lead to a build-up of AAT in the liver. This build-up may cause liver disease linked to AATD.

2. What is the Service for?

- **Detects** 14 variants in the *SERPINA1* gene linked to AATD. These 14 variants explain 95% of AATD cases
- **Determines** if your genetics increases your risk of developing lung and/or liver disease linked to AATD compared to the general population

Read information on the **14 genetic variants**

Read information on the **risk categories** used by the Service

View the **Package Insert** for more information about special considerations for testing, and clinical and analytical performance of the Service

3. How accurate is the Service?

This Service is at least 99% accurate. For more details on the performance of the Service, please refer to the **Package Insert**

4. My report says I have "2 variants" in the *SERPINA1* gene linked to AATD. How did I get these variants?

Results for this Service will show that there is no, 1, or 2 variants in the *SERPINA1* gene. Based on your results, you have 2 variants in the *SERPINA1* gene linked to AATD.

You inherited 1 variant from each of your parents. So, each of your parents must have at least 1 variant. Talk to a healthcare professional about family testing.

5. My report says I have "2 variants, PI*Z and PI*Z". What are some next steps I can take?

1. **Share your results report with a healthcare professional.**
   Healthcare professionals can answer questions you may have about your results, risk, and how they may apply to your health. They should know you were tested for AATD. You should also inform a healthcare professional if you:
   - Have symptoms of lung or liver disease (see page 8 for more information)
   - Have a personal or family history of lung or liver disease
   - Are feeling anxious, uncertain, or concerned about your genetic result or risk
   - Have questions about any risk factors
2. **Consider sharing this report with your family members.**
   Since you share genetics with biological family members, your result may be important for their health as well. Talk to a healthcare professional about family testing.
3. **Use the links below to follow through on these next steps:**
   - To find a doctor with experience in AATD from the Alpha-1 Foundation's Clinical Resource Centers, visit **http://www.alpha1.org/alphas-friends-family/resources/find-an-alpha-1-specialist**
   - To find a healthcare professional near you with experience testing for AATD, please visit **www.AlphaFindADoctor.com**
   - To find a genetic counselor, visit **https://findageneticcounselor.nsgc.org/**

For information about more resources, see page 14.

6. What does "at increased risk of developing lung disease linked to AATD compared to the general population" mean?

This means your chance of developing lung disease linked to AATD is higher than that of the general population. It does not mean that you will develop lung disease.

In addition to your genetic result, there are other non-genetic factors that can impact your risk of developing lung disease, eg, smoking and environmental pollution at work or around your home.

It is important to share this results report with a healthcare professional. Healthcare professionals can answer questions you may have about your results, risk, and how they may apply to your health. They should know you were tested for AATD.

7. What does "at slightly increased risk of developing liver disease linked to AATD compared to the general population" mean?

This means your chance of developing liver disease linked to AATD is slightly higher than that of the general population. It does not mean that you will develop liver disease.

In addition to your genetic result, there are other non-genetic factors that can impact your risk of developing liver disease, eg, alcohol consumption.

It is important to share this results report with a healthcare professional. Healthcare professionals can answer questions you may have about your results, risk, and how they may apply to your health. They should know you were tested for AATD.

8. Can AATD be treated?

Yes. While there is no cure for AATD, it is important to know that treatment options may be available.

Augmentation therapy is a treatment option for lung disease linked to severe AATD. It raises the level of AAT protein in the blood. Also, it is important for people with AATD to avoid smoking.

There are no current specific treatment options for liver disease linked to AATD. The treatment options are the same ones that are used for liver diseases in general. People with liver disease linked to AATD should avoid alcohol consumption.

9. What are the limitations of the Service?

- It is not a substitute for an appointment with a healthcare professional. We strongly recommend you consult with a healthcare professional if you have any questions or concerns about your result or health
- It does not diagnose any disease or condition. Only a healthcare professional can diagnose a disease or condition
- It does not determine if you have or will develop lung and/or liver disease linked to AATD during your lifetime
- It cannot be used to make healthcare decisions. It does not tell you anything about your overall health. Only a healthcare professional can help you with healthcare decisions
- It detects 14 variants in the *SERPINA1* gene linked to AATD. These 14 variants explain 95% of AATD cases. It does not detect all possible variants linked to AATD
- There may be other, non-genetic factors that affect your risk. The Service does not determine your overall risk of developing lung and/or liver disease
- Other companies that offer AATD testing may detect different or fewer variants linked to AATD. You may get different genetic results using a test from a different company
- The laboratory may be unable to process every person's sample. If this happens, you will receive a notice and another AlphaID™ At Home Saliva Collection Kit so that you can provide a new sample to the laboratory

10. How can your genetic result affect your family?

AATD is a genetic condition. It is passed from your biological parents to you. You can pass AATD on to your children. Everyone has 2 copies of the ***SERPINA1*** gene. One copy of this gene was passed to you from each of your parents. Since you share genetics with your family, your result may be important for their health, too.

You have **2 variants** in the ***SERPINA1*** gene linked to AATD. This means:

- You inherited 1 variant from each of your parents
- You will pass 1 of your 2 variants to your children
- If your child's other biological parent has a *SERPINA1* variant, your child could inherit two variants, one from each parent. This may put them at risk of developing lung and/or liver disease linked to AATD

The American Thoracic Society and the COPD Foundation recommend testing for variants linked to AATD in family members—parents, siblings, and children—of a person with AATD.

11. My report says AATD is most common in people of European descent. What if I'm not of European descent?

AATD occurs in people of all ethnicities worldwide. However, it is most common in people of European descent, because they are more likely to have variants in the *SERPINA1* gene. AATD affects about 1 in 1,500 to 3,500 people of European descent.

People of non-European descent can still have variants in the *SERPINA1* gene that increase their risk of developing lung and/or liver disease linked to AATD. This means even if you are not of European descent, your genetic risk report still applies to you.

12. Are there resources available for me to find a healthcare professional experienced in AATD?

Here are 3 resources to connect you with a healthcare professional who has experience in AATD:

- The Alpha-1 Foundation has Clinical Resource Centers, where you can find a healthcare professional who is knowledgeable about AATD. Please visit **www.alpha1.org/alphas-friends-family/resources/find-an-alpha-1-specialist**
- To find a healthcare professional near you with experience testing for AATD, please visit **www.AlphaFindADoctor.com**
- To find a genetic counselor, please visit **https://findageneticcounselor.nsgc.org/**

13. Where can I learn more about AATD?

The following organizations and advocacy groups can provide resources, education, and/or support for AATD:

Alpha-1 Foundation

The Alpha-1 Foundation is the only national organization dedicated to developing a cure for alpha-1 antitrypsin deficiency and improving the quality of life for patients and their families. Acting as a patient advocate, the foundation helps the AATD community better connect to one another – strengthening the support and care networks they need. **https://www.alpha1.org** | (877) 228-7321 ext. 321

AlphaNet

AlphaNet is a not-for-profit organization providing innovative health management services and customized care to the AATD community via care coordinators who are AATD patients themselves.
https://www.alphanet.org | (800) 577-2638

COPD Foundation

The COPD Foundation is a not-for-profit organization established to speed innovations that will make treatments more effective and affordable, undertake initiatives that result in expanded services for COPD patients, and improve the lives of patients with COPD.
https://www.copdfoundation.org

American Liver Foundation

American Liver Foundation provides community education programs, funds research, and supports advocacy related to liver disease.
https://liverfoundation.org/

MedlinePlus

The National Institutes of Health provide consumer-friendly information about the effects of genetic variation on human health.
https://medlineplus.gov/genetics/condition/alpha-1-antitrypsin-deficiency

Thank you for taking this important step to find out about your genetic health risk linked to AATD.

Please share your results with a healthcare professional. Healthcare professionals can answer questions you may have about your results, risk, and how they may apply to your health now and in the future.

Glossary of Terms

Here are the common key terms from your results report.

Alpha-1 antitrypsin (AAT)

Alpha-1 antitrypsin (AL-fa / one / an-tee-TRIP-sin) is a protein that is produced by instructions from the *SERPINA1* gene. The AAT protein is released by the liver. The blood transports it to the lungs. Having low levels of the AAT protein can leave your lungs vulnerable to developing lung disease linked to alpha-1 antitrypsin deficiency.

Alpha-1 antitrypsin deficiency (AATD)

Alpha-1 antitrypsin deficiency is a genetic condition that is caused by variants in the *SERPINA1* gene. This leads to having low levels of the AAT protein. The AAT protein plays a critical role in safeguarding the lungs. Low levels of the AAT protein leave the lungs vulnerable to developing lung disease linked to AATD.

This condition can also lead to a build-up of AAT in the liver. This build-up may cause liver disease linked to AATD.

AlphaID™ At Home Genetic Health Risk Service

An at-home genetic testing Service provided by Grifols. The AlphaID™ At Home Genetic Health Risk Service provides consumers with their risk of developing lung and/or liver disease linked to alpha-1 antitrypsin deficiency by detecting 14 variants in the *SERPINA1* gene linked to AATD. These 14 variants explain 95% of AATD cases. Scientific information on the **14 genetic variants is found here.** Definitions of each potential **risk category are found here.**

AlphaID™ At Home Genetic Health Risk Service Report

This report explains your genetic result and interprets a risk for developing lung and/or liver disease linked to alpha-1 antitrypsin deficiency compared to the general population. General population is defined as all adults who live in the United States.

Bronchiectasis

Bronchiectasis is caused by the airways of the lungs becoming damaged and widened. It is one of the common lung diseases or complications linked to AATD.

Chronic bronchitis

Chronic bronchitis involves inflammation and swelling of the lining of the airways. This swelling leads to narrowing and obstruction that can result in a daily cough. The inflammation stimulates production of mucus. Mucus can cause further blockage of the airways. It is one of the common lung diseases or complications linked to AATD.

Chronic hepatitis

Chronic hepatitis is inflammation of the liver that lasts at least 6 months. It is one of the common liver diseases or complications linked to AATD.

Chronic obstructive pulmonary disease (COPD)

COPD is a serious lung disease characterized by emphysema, persistent airflow obstruction, and/or chronic bronchitis. It is one of the common lung diseases or complications linked to AATD.

Cirrhosis

Cirrhosis is a condition in which the liver does not function properly due to long-term damage. This damage replaces normal liver tissue with scar tissue. It is one of the common liver diseases or complications linked to AATD.

DNA

DNA is like an instruction manual with the information needed for your body to work. It is also responsible for all the physical traits that are passed down to you from your biological parents. Information is stored in DNA like a code.

Emphysema

Emphysema is a condition in which the walls of the air sacs at the bottom of the lungs (called “alveoli”) are degraded, causing loss of the lung tissue, enlargement, and breathlessness. It is one of the common lung diseases or complications linked to AATD.

Ethnicity

Ethnicity is a social construct that defines groups of people with common ancestry.

Gene

A gene is a basic unit of genetic information. Genes are made up of DNA. Genes give your body specific instructions for making proteins. Proteins are the molecules that run your body. You have 2 copies of your genes. One copy was passed to you from each of your biological parents.

Genetic counselors

Genetic counselors are healthcare professionals who have education in genetics and counseling. They help people understand and make decisions about their genetic health. They interpret genetic test results and guide and support people seeking more information about their genetic backgrounds.

Genetic inheritance

Genetic inheritance refers to the passing of genes down from one generation to the next (eg, from the mother to the child). Each person has two copies of every gene. One copy from the biological mother and one from the biological father.

Genetic result

Your genetic result, also called genotype, contains the number and names of the variants you have out of the 14 variants tested by the AlphaID™ At Home Genetic Health Risk Service. A person's genotype includes two copies of the *SERPINA1* gene. The normal version of the *SERPINA1* gene (without any variants) is PI*M.

- A person with genetic result "no variants" has the genotype PI*M/PI*M (PI*MM).
- A person with genetic result "1 variant", for example PI*Z, has the genotype PI*M/PI*Z (PI*MZ).
- A person with genetic result "2 variants", for example PI*S and PI*Z, has the genotype PI*S/PI*Z (PI*SZ).

Jaundice

Jaundice is a yellowish discoloration of the white part of the eyes and skin due to high bilirubin levels. It is one of the common liver diseases or complications linked to AATD.

Liver disease

Liver disease is any problem in the liver that prevents the liver from working properly. The common liver diseases or complications linked to AATD include cirrhosis, jaundice, hepatic enzyme elevations, chronic hepatitis, and liver scarring (fibrosis).

Liver scarring (fibrosis)

Liver scarring occurs when repetitive or long-lasting injury or inflammation causes excessive amounts of scar tissue. It is one of the common liver diseases or complications linked to AATD.

Lung disease

Lung disease is any problem in the lungs that prevents the lungs from working properly. The common lung diseases or complications linked to AATD include chronic obstructive pulmonary disease (COPD), emphysema, chronic bronchitis, and bronchiectasis.

Risk factor

Something that increases a person's chance of developing a health condition is a risk factor. For example, smoking is a risk factor for alpha-1 antitrypsin deficiency. People's lifestyle choices and environmental factors can affect their risk of developing lung and/or liver disease linked to alpha-1 antitrypsin deficiency. Common risk factors include smoking, alcohol consumption, environmental pollution exposure at work and home, obesity, personal or family history of lung and/or liver disease, lung infections, and liver infections.

SERPINA1 gene

The *SERPINA1* gene provides instructions to the body for making AAT protein in the liver. Variants of the *SERPINA1* gene were named by the prefix PI* (protease inhibitor*), which serves as another name for the *SERPINA1* gene. Using this naming system, one of the most common variants linked to alpha-1 antitrypsin deficiency is PI*Z.

Variant

A permanent change or difference in the DNA sequence of a gene among individuals is called a variant. Most variants are harmless, but others are linked to an increased risk, compared to the general population, for a variety of health conditions.

