TEST

Alpha-1 antitrypsin blood test

A blood test that measures alpha-1 antitrypsin protein to help identify an inherited deficiency.

WHAT IT MEASURES
Amount of alpha-1 antitrypsin protein in blood

SAMPLE
Blood

FASTING NEEDED?
No

HOW LONG IT TAKES
A blood draw usually takes under ten minutes

RESULTS IN
Several days; varies by laboratory

USED FOR
Screening and diagnosing inherited alpha-1 antitrypsin deficiency

MAIN LIMITATION
Inflammation can raise levels and mask deficiency

Overview

The alpha-1 antitrypsin blood test measures alpha-1 antitrypsin, or AAT. This protein is made mainly in the liver and helps protect lung tissue from enzymes released during inflammation.

Some people inherit changes in the `SERPINA1` gene that cause a low AAT level or an abnormal protein. This is called alpha-1 antitrypsin deficiency. It can increase the chance of lung or liver problems, but results cannot predict whether these will develop.

A quantitative test measures the amount of AAT. If the result is low or does not fit your medical history, additional tests may identify the protein type or gene variant involved.

Why the test is ordered

The test may be ordered when lung or liver disease could be related to inherited AAT deficiency or to screen relatives of someone with a confirmed deficiency.

Reasons include:

The test does not by itself explain symptoms or confirm the cause of lung or liver damage.

- Chronic obstructive pulmonary disease, or COPD
- Emphysema, especially at a younger age or without substantial smoking exposure
- Unexplained bronchiectasis
- Unexplained long-term liver disease
- A family history of AAT deficiency
- Rare associated conditions, such as necrotizing panniculitis

Who should have the test

Clinical guidance supports offering testing to people with COPD or unexplained chronic liver disease. It may also be considered for unexplained bronchiectasis or certain uncommon inflammatory conditions.

Parents, siblings, children, and other relatives of someone with a confirmed harmful `SERPINA1` variant may be offered testing. Genetic counselling can help explain how inherited results may affect relatives.

Routine population-wide testing is not standard everywhere. Whether testing is appropriate depends on your health history, family history, previous results, and local practice.

How to prepare

You do not usually need to fast. You can generally eat, drink water, and have coffee normally unless another test at the same appointment requires fasting.

Tell the healthcare professional if you:

These factors may affect the result or its interpretation. Do not stop prescribed medicines unless the clinician responsible for them tells you to.

- Have an infection, fever, recent injury, or active inflammatory condition
- Are pregnant
- Take an estrogen-containing medicine, including some contraceptives
- Recently received plasma or another plasma-containing blood product
- Have severe liver disease, kidney disease, or abnormal protein loss

What happens during the test

A healthcare professional cleans the skin, usually inside your elbow, and inserts a needle into a vein. A small amount of blood is collected into a tube. The needle is removed and pressure is applied.

The blood draw usually takes only a few minutes. If phenotype or genetic testing is needed, the same sample may be used, although another sample is sometimes required.

Understanding your results

Your report will show an AAT concentration and the laboratory's reference range. Interpretation varies by testing method and clinical context.

A low AAT level may suggest inherited deficiency. It can also occur if the liver is not making proteins normally or protein is being lost through the kidneys or digestive tract. Phenotype or genetic testing commonly follows.

A result within the laboratory range makes severe deficiency less likely but does not always rule it out. Infection or inflammation can temporarily raise a low baseline level, and some uncommon variants produce a normal amount of protein that does not work normally.

A high AAT level is usually nonspecific. AAT rises in response to inflammation, infection, pregnancy, or estrogen exposure. A high result does not identify the cause or location of inflammation.

Follow-up tests may include:

- Phenotyping to identify forms of the AAT protein
- Genotyping to look for selected `SERPINA1` variants
- Gene sequencing to search more broadly for uncommon variants
- Liver and lung tests when clinically appropriate

What affects the results

Inflammation is the main source of misleading results. Infection, injury, or inflammatory illness can raise AAT and hide an underlying deficiency. A C-reactive protein test may be measured alongside AAT to check for inflammation.

Pregnancy and estrogen-containing medicines can raise AAT. Severe liver dysfunction may

lower it, while kidney or intestinal conditions that cause protein loss can produce a low result without inherited deficiency.

A recent plasma-containing transfusion can temporarily affect the protein level or phenotype. Tell the testing service when it occurred. Standard genetic testing is not usually affected, although previous stem-cell or bone-marrow transplantation can complicate genetic testing from blood.

Meals, coffee, time of day, and ordinary exercise have little direct effect. Smoking does not usually cause a falsely low level, but it increases lung damage and may reduce AAT's protective effect.

Laboratories use different methods, units, and reference ranges, so results from different laboratories may not be directly comparable.

Accuracy and limitations

A quantitative test identifies many people with significant deficiency, but inflammation, pregnancy, or estrogen can raise the level and mask it. The test measures the amount of protein, not whether it works normally.

A low result does not always indicate inherited deficiency because liver failure or abnormal protein loss can have the same effect. Confirmatory testing helps distinguish these causes.

Phenotyping identifies several common protein forms but may not classify rare variants or variants producing no detectable protein. Targeted genotyping finds only the variants included in the test, often the common S and Z variants.

Broader `SERPINA1` sequencing, sometimes with testing for gene deletions, may be appropriate when the concentration, phenotype, genotype, and medical history do not agree. No single test predicts future lung or liver health.

Risks and complications

The main risks are brief discomfort, a small bruise, and feeling light-headed during or after the blood draw. Ongoing bleeding or infection at the needle site is uncommon.

What happens next

A normal result that fits your health history may need no further testing. If the test was done

during an infection or inflammatory episode, it may be repeated after recovery.

A low, borderline, or unexpected result should be discussed at a follow-up appointment. Phenotype or genetic testing may be arranged, and relatives may be offered information about testing. Lung function or liver blood tests may be considered based on your history.

Seek prompt medical advice for new jaundice, worsening abdominal swelling, coughing up blood, or a marked change in breathing. Call emergency services for severe breathing difficulty, blue or grey lips, fainting, confusion, or vomiting blood. Do not wait for a routine results appointment.

Common questions

Can I drink coffee before the test?

Yes. Coffee does not usually affect AAT, and fasting is not normally required. Follow any instructions for other tests being collected at the same time.

Does the time of day matter?

Usually not. AAT does not have a clinically important daily pattern requiring morning testing.

Can I exercise beforehand?

Ordinary exercise is unlikely to affect the result. Mention recent major injury, surgery, or unusually intense exercise that caused significant inflammation or muscle damage.

Can a cold affect my result?

Yes. Infection can raise AAT and hide a low baseline level. The test may need to be repeated after recovery.

Is this a genetic test?

Not usually. The first test measures AAT protein in blood. Phenotype or genetic testing may follow if the result is low, borderline, or inconsistent with your history.

Will one normal result rule out alpha-1 antitrypsin deficiency?

Not always. Inflammation, pregnancy, estrogen medicines, and uncommon variants can make a normal concentration misleading. Further testing may be appropriate if clinical suspicion remains.

Sources

- Sandhaus RA, Turino G, Brantly ML, et al. "The Diagnosis and Management of Alpha-1 Antitrypsin Deficiency in the Adult." Journal of the COPD Foundation . 2016;3(3):668-682.
- American Thoracic Society and European Respiratory Society. "Standards for the Diagnosis and Management of Individuals with Alpha-1 Antitrypsin Deficiency." American Journal of Respiratory and Critical Care Medicine . 2003;168(7):818-900.
- Miravitlles M, Dirksen A, Ferrarotti I, et al. "European Respiratory Society Statement: Diagnosis and Treatment of Pulmonary Disease in Alpha-1 Antitrypsin Deficiency." European Respiratory Journal . 2017;50(5):1700610.
- Stoller JK, Hupertz V, Aboussouan LS. "Alpha-1 Antitrypsin Deficiency." GeneReviews , University of Washington, Seattle.
- ARUP Consult. "Alpha-1 Antitrypsin Deficiency." Test-selection and interpretation guidance.
- MedlinePlus Medical Test. "Alpha-1 Antitrypsin Test." U.S. National Library of Medicine.