Alpha-1 Antitrypsin

Does this test have other names?

Alpha-1-antiprotease deficiency, alpha-1-antiproteinase inhibitor deficiency, AAT deficiency, alpha-1-antitrypsin (A-1AT) deficiency

What is this test?

This is a type of blood test. It helps find out if you have a genetic disorder called alpha-1 antitrypsin deficiency. This disorder is linked to abnormally low levels or a lack of alpha-1 antitrypsin (AAT) protein in the blood. The disorder can cause liver and lung diseases. These health problems may show up much earlier than they normally would.

AAT is a protein made in the liver that helps your lungs work normally. The test finds out whether you have normal or damaged copies of a gene that makes this protein. If you have AAT deficiency, you have two damaged copies of this gene. This makes it more likely that you will develop serious lung or liver problems.

Although it's helpful to know whether you have this genetic disorder, a positive test result may affect your mental health and your health insurance (although laws were passed to protect against discrimination for health insurance based on having a genetic condition). For this reason, some groups, including the nonprofit Alpha-1 Foundation, advise talking with a genetic counselor before getting a medical test that will stay in your health records. Contact a health professional who specializes in advising patients about genetic diseases.

Why do I need this test?

The only way to find out whether you have this genetic disorder is to get tested.

Your healthcare provider may order this test if you have these problems:

- Chronic wheezing
- Asthma that can't be controlled through aggressive treatment
- Shortness of breath during mild exercise
- Rapid heartbeat when you stand up
- Vision problems
- Chronic bronchitis
- Bumpy or itchy skin
- Chronic obstructive pulmonary disease (COPD)
- Emphysema, a serious, progressive lung disease that causes breathing problems
- Bronchiectasis, a lung problem that causes chronic dilation of the airways
- Blood in your vomit
- Swelling in your abdomen or legs
- Unexplained liver problems, especially if they appear in childhood
- Unintentional weight loss

Infants who have yellow skin, a sign of jaundice, may also be tested.

Your healthcare provider may also recommend the test if someone in your family has a history of the above conditions.

What other tests might I have along with this test?

Your healthcare provider may recommend other tests to rule out other conditions:

- Pulmonary function testing. A series of tests to see how well your lungs work.
- Chest radiography. X-ray of your lungs.
- HRCT (high-resolution CT) and densitometry. More detailed X-rays of your lungs and tests to measure any lung and liver damage.
- Liver function tests. A series of blood tests to see how well your liver works.
- Abdominal ultrasound. A test that uses sound waves to find evidence of liver damage.
- Liver biopsy. A procedure that involves taking a tiny sample of your liver to look for disease.

Your healthcare provider may do a series of tests on the serum in your blood to find the damaged genes. These tests include:

- Serum AAT concentrations. Tests on a blood sample to look for the AAT protein.
- Serum protein electrophoresis with thin-layer isoelectric focusing. A more accurate test for the gene.
- Genetic test. You may consider getting a genetic test before getting a blood test to look for the amount of AAT protein you have. The genetic test can be done with a blood sample or a swab from inside your mouth.

What do my test results mean?

Test results may vary depending on your age, gender, health history, and other things. Your test results may be different depending on the lab used. They may not mean you have a problem. Ask your healthcare provider what your test results mean for you.

Labs may vary in their results. But a typical normal result will be between 75 and 150 milligrams per deciliter (mg/dL), depending on how the results were done. If your levels are too low, it may be a sign that you have one damaged gene, which means you are a carrier, or two damaged genes, which means you have AAT deficiency. Talk with your healthcare provider about what your test results mean.

How is this test done?

The test is done with a blood sample. A needle is used to draw blood from a vein in your arm or hand.

Does this test pose any risks?

Having a blood test with a needle carries some risks. These include bleeding, infection, bruising, and feeling lightheaded. When the needle pricks your arm or hand, you may feel a slight sting or pain. Afterward, the site may be sore.

Although the physical risks of the test are slight, it may be emotionally upsetting to find out you have a genetic risk for certain diseases. Because it may reveal a chronic health condition, the test may also affect your coverage for life insurance. For these reasons, experts recommend seeking genetic counseling.

What might affect my test results?

If the test is done correctly, the results should be accurate. But there is always the chance of a false abnormal result. You can also have a low ATT level from certain conditions such as kidney disease, malnutrition, and some cancers. Or you can have a temporary increase in your ATT level from an infection, stress, pregnancy, or the use of some medicines, such as birth control pills. This can lead to a normal level when the level may actually be low.

How do I get ready for this test?

You don't have to prepare for this test.