KAISER PERMANENTE®

INFORMATION SHEET

Alpha-1 Antitrypsin Deficiency

Basic Facts About Alpha-1 Antitrypsin Deficiency

- Alpha-1 antitrypsin (AAT) is a protein made in the liver.
- AAT helps protect the lungs from damage due to infections or inhaled irritants (ex: smoking).
- A person with AAT deficiency (AATD) has a higher chance for liver disease and lung disease.
- About 1 in 3000 people has AATD. It is more common in people with European ancestry.

What causes AATD?

AATD is inherited. It is caused by changes in the AAT gene (also called SERPINA1).

The most common version of the AAT gene is called M. Most people have two copies of M ("MM") and make normal AAT. There are more than 150 AAT variants (mutations) that change how the protein is made. These changes can lead to less AAT in the blood and more AAT in the liver.

A person with AATD inherits two variants in the AAT gene, one from each parent. The most common AAT variants are called S and Z. These two variants lower the amount of AAT in the blood by a little (S variant) or a lot (Z variant). The chance for health problems depends on which two variants a person has. It also depends on a person's exposure to things that can damage the liver or lungs. Some people with two variants have healthy lives without any lung or liver problems.

Possible Test Results for AATD:

When genetic testing shows:

ZZ – There is a higher chance for liver disease at any age, even in childhood. Alcohol use or hepatitis can make liver failure more likely to happen. There is also a higher chance for lung disease, even in non-smokers. Lung disease rarely happens before age 40. Smoking greatly increases the chance for lung disease.

SZ – There is a higher chance for adult lung disease, especially if you smoke. There is also a slightly higher chance for liver disease.

SS – The chance for liver disease or lung disease is only slightly increased. Exposures can increase the chance.

MS or MZ - Almost 1 in 14 people in North America have one copy of S or Z. A person with only one AAT variant is a carrier, but does not have AATD. These people are unlikely to develop liver or lung disease unless they are exposed to smoking, heavy alcohol use, or other risk factors.

Sometimes the amount of AAT protein is measured by taking a blood sample. Carriers (people with just one variant) often have AAT levels a little lower than normal and this does not cause disease. A person with AATD who is at risk of liver or lung disease has much lower AAT levels than normal. More testing may be ordered to look for rare variants if AAT is extremely low in a carrier.

Who should be tested for AATD?

There is no reason for a doctor to test someone for AATD unless they have liver or lung disease, or a close relative with AATD.

It is also uncommon to test children for AATD unless they have liver disease. The chance of liver disease in a child is low unless there are other children in the family with liver disease.

How is AATD treated?

There is usually no special treatment needed for AATD. A person with AATD who develops lung disease or liver disease will be treated based on guidelines for those conditions. Someone who does not have symptoms should avoid exposure to cigarette smoke and other lung irritants. They should also get vaccines that protect against infections in the liver and lungs.

RESOURCES:

Alpha-1 Foundation
3300 Ponce de Leon Blvd. Coral Gables, Florida 33134
Main: (305) 567-9888
Toll Free: (877) 2-CURE-A1 or (877) 228-7321
Email: info@alpha1.org
www.alpha1.org

MedlinePlus
Alpha-1 antitrypsin deficiency