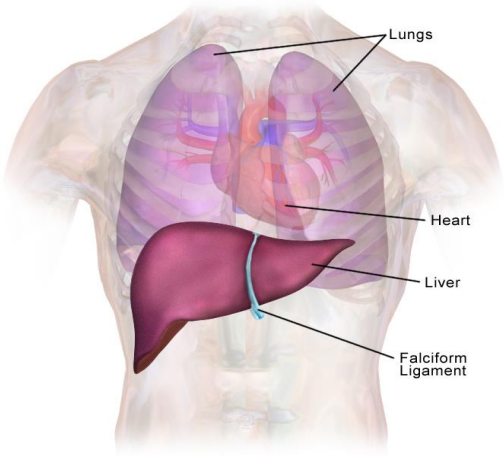
Fact Sheet - Liver Health & Alpha-1 Antitrypsin Deficiency

Background

Alpha-1 antitrypsin deficiency (A1AD) is a common serious genetic disorder, passed on from parents to their children. A1AD is found in most countries but usually in individuals with a European background (e.g. Scandinavian, British, French, Spanish). Although A1AD occurs in one in nine people in Australia and can cause serious lung and liver disease, the community and doctors are poorly informed about A1AD. The Alpha-1 Organisation Australia (A1OA) is working hard to change this situation.



Placement of the Liver

The Role of the Liver

The liver is a very important organ. It cleans your blood, stores sugars, fats and vitamins and nutrients from food. The liver breaks down alcohol, medication and toxic substances that could harm your body. Liver disease means that the liver isn't working as well as it should.

What Causes Liver Disease in A1AD?

Liver problems can appear in infants, children and adults. Alpha-1 antitrypsin (AAT) is a protein made mostly by the liver. AAT is meant to leave the liver and circulate around the body to protect other organs. Liver disease can happen if AAT becomes stuck in the liver due to a genetic change that can change the shape of the AAT and it becomes stuck in the liver.

Liver disease from trapped AAT is the second most common health problem associated with A1AD (the main problem is with the lungs). When the protein becomes stuck it is called a polymer. The liver can't break down the polymer and a build-up can cause liver damage, fibrosis and serious scarring called cirrhosis. In some people the liver is so damaged that liver cancer can occur.

There are over 150 A1AD gene mutations. The mutation called ZZ is most commonly associated with liver disease as the Z gene is "misfolded" and its shape causes it to become stuck.

Scientists don't know why some individuals have A1AD liver disease and others do not. Currently there is no way to prevent the protein from building up in the liver but clinical trials are underway and a cure may be possible within the next 10 years.

Children

Most children with ZZ genotype would have abnormal liver function test results but results return to normal during their childhood. Some infants will have jaundice for a lot longer than other infants which is an indication that the AAT may be trapped in their liver. About 10% of ZZ will experience liver problems. Some ZZ children may be born with liver disease or develop it later. Unfortunately, out of these children, 2% will have liver failure and will need a liver transplant in the first two years of life.

Adults

Older ZZ adults are likely to have liver scaring than younger adults but may have normal liver function test results until late in life (e.g. over 60 years of age). It is estimated that around 30% of ZZ adults will develop liver disease.

Some MZ adults will have liver disease although they are thought to be at lower risk of disease than ZZs. It is thought that previous damage to the liver may predispose MZ individuals to A1AD liver disease.

Liver-affected individuals with A1AD can benefit from a lifestyle that supports liver health.

What are the Symptoms of Disease?

- Eyes and skin turning yellow (jaundice)
- Swelling on the abdomen (ascites) or legs
- Vomiting blood or passing blood in the stool
- Abnormal itching (pruritis)

Treatment for Liver Disease

There is no specific treatment to prevent liver disease or to treat it once diagnosed. Rather doctors will give supportive care and manage the health problems being experienced. In severe cases treatment is liver transplantation. Augmentation therapy does not help A1AD liver disease.

Activities to Support Liver Health

Things that can harm the liver include viruses e.g. hepatitis A, B or C and substances such as alcohol. It is advisable to be immunised against hepatitis A and B and to limit alcohol. Alphas should exercise and eat a healthy diet, avoid carbohydrates and sugar which can lead to a “fatty liver” which can also cause liver disease if not reversed through healthy eating.

What Tests Can Find Liver Disease?

Liver disease can be found by blood tests e.g. liver function tests and by scans e.g. ultrasound or a Fibroscan (measures the stiffness of your liver). A liver biopsy is rarely needed although may be required in severe disease to assist with diagnosis. It is advisable to have regular (e.g. annual) liver tests. People over 50 who have worsening cirrhosis are at risk of liver cancer so periodic CT scans of the liver are recommended.

Who Should be Tested for Alpha-1?

- Anyone with unexplained liver disease including newborns, children and adults
- Relatives of individuals diagnosed with A1AD
- People with a family history of liver disease
- Everyone with a diagnosis of “adult asthma”, chronic obstructive pulmonary disease (COPD), chronic bronchitis and early onset emphysema (e.g. by 35-40 years of age in smokers; 55-60 in non-smokers).

Need More Information or Support?

Contact Alpha-1 Organisation Australia : email contactus.a1oa@gmail.com

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This brochure is one in a series of information sheets produced by the Alpha-1 Organisation Australia (A1OA). This information is designed to be a guide only and does not replace advice given by your health professional. Any treatment information or brand names are correct at the time of printing. If the information raises concerns or if you have further questions please consult your doctor.