Alpha-1 Antitrypsin Deficiency

Patient and family information, brought to you by the Education Committee of APSA

Overview - "What is it?"

Alpha-1 antitrypsin deficiency (AATD) may cause liver problems in children. It is passed down from parents to the child (inherited). Alpha-1 antitrypsin is a protein made by the liver that goes into the bloodstream to help protect other tissues from damage by certain chemicals. (See Figure 1) In AATD, there is abnormality of the alpha-1 antitrypsin protein, the protein builds up in the liver and causes damage to the liver. The effect of AATD is different in all patients. Only about 1 in 20 babies born with AATD will get liver disease. Sometimes the damage to the liver is bad enough that a liver transplant is needed. Because alpha-1-anti-trypsin protein protects some body parts from harm by certain chemicals, defective protein can result in damage to organs.

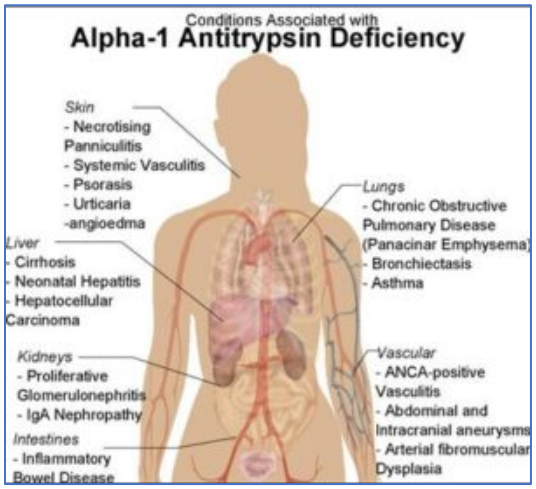

Figure 1: Possible diseases associated with AATD.
Image credit: wikipedia

This condition can affect 1 person in every 3,000 to 5,000 people or about 3.4 million people in the world. White people (Caucasian) are most frequently affected. Boys and girls are affected in equal numbers.

Signs and Symptoms - "What symptoms will my child have?"

Early on, newborn babies with AATD may have yellowing of the white of the eyes and the skin (jaundice), swelling of the belly, poor growth, poor appetite. Many babies and children with AATD will have NO symptoms.

Older children and adults may feel tired and weak. Older adults may begin to have difficulty breathing as smoke and chemicals in the environment begin to damage the lungs since they cannot be protected by the abnormal alpha-1 antitrypsin protein. Other body systems that can have problems include skin (itching, hives), liver, kidneys and blood vessels.

Diagnosis - "What tests are done to find out what my child has?"

- Physical examination by a doctor.
- Labs and tests: In a baby or child with jaundice, the doctor will examine the belly to see whether the liver is big or if there is fluid in the belly. Blood tests will check the liver function and to check whether the alpha-1 antitrypsin protein is normal. A genetic test can check the genes that make the alpha-1 antitrypsin protein.
- Sometimes the doctor will order an ultrasound test to look at the liver and gallbladder. This test does not cause any pain and uses soundwaves to show a picture of the liver and gallbladder.
- Other conditions that mimic AATD: Most babies have some jaundice during the first few days of life. Some babies with jaundice need to be under special lights to make the jaundice go away. Most babies with jaundice do not have AATD. There are other problems that cause jaundice such as biliary atresia, hepatitis, cystic fibrosis, choledochal cyst (cyst of the bile ducts), liver infections and viruses. Your doctor will rule some tests to make sure check for these conditions.

Treatment - "What will be done to make my child better?"

Medicine: There is no cure for AATD. The treatment is aimed at making the child's symptoms better and to limit the damage to the liver. Your doctor might suggest a special diet and avoid medicines that may damage the liver.

Surgery: The need for surgery in babies and children with AATD is rare. Sometimes children will undergo a liver biopsy to confirm the diagnosis. A biopsy is when a piece of liver is taken and studied to see what is wrong with it. In rare cases, the liver damage could be very serious, and the child will need a liver transplant.

- **Transplant risks/benefits:** A person needs his or her liver to work well. If the liver is severely damaged by AATD, then a liver transplant is needed. When a person gets a liver transplant, he or she will need to be on many medicines for the rest of their life to protect against infection and rejection (the body fighting against the new liver).

Home Care - "What do I need to do once my child goes home?"

Diet: Good nutrition is very important so the child continues to grow. You may meet with a pediatric dietician to review the best foods to give your child for adequate growth.

Activity: Unless your child has surgery, there are no limits to activity.

Medicines: Your child may be given medicines to help with any of the symptoms of AATD, such as medicine for fluid build-up or for itching. Your child will likely need vitamins.

What to call the doctor for: Call your pediatrician for any bleeding, enlarging abdomen, decreased energy, decreased appetite or severe itching.

Follow-up care: You will need to see your pediatrician for regular exams and bloodwork. It is important have shots (vaccinations) as directed by the doctor.

Long Term Outcomes - "Are there future conditions to worry about?"

People with AATD may develop lung problems later in life and be diagnosed with "emphysema" (chronic damage to the lungs). Children and adults with AATD should NOT smoke or be exposed to smoke and other environmental toxins. Smoke will worsen the damage to the lungs.

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