A guide for parents of children diagnosed as having Alpha-1 Antitrypsin Deficiency
Before we begin, you must know that nothing written here should be taken as a substitute for what your Doctor or other Consultant may say. This should be read in conjunction with our “Newly Diagnosed” leaflet to give a better understanding of the condition.

You have been told by your doctor that your child has Alpha-1 Antitrypsin Deficiency (AATD) and that there may be problems with their liver.

While this is distressing please understand that it is NOT a life sentence. With care and understanding your child can still have a long and full life.

This leaflet attempts to explain and support, using the minimum of technical terms, what your doctor or hospital consultant may have discussed with you.

Your Doctor, or Consultant, should have told you that your child’s liver is either producing insufficient or defective alpha-1 antitrypsin protein.

You may already be aware that you and/or the other parent of your child also suffers from Alpha-1 Antitrypsin Deficiency.

If you have not yet been tested then it is very important that you are. You should request the test from your GP as soon as possible.
Understanding the Terminology:

Alpha-1 Antitrypsin Deficiency is a condition where people have lower than normal amounts of alpha-1 antitrypsin. In much the same way as hair and eye colour is passed from parent to child, so alpha-1 antitrypsin deficiency is also passed from parent to child. Each child gets one gene from their mother and the other from their father.

Alpha-1 is a protein that is produced in the liver and travels in the blood stream to the lungs where it protects the lining of the lungs.

The gene in the liver that produces alpha-1 antitrypsin (AAT) is known as a Protease inhibitor (Pi). A non affected person is known as PiMM (‘M’ meaning normal), where the liver produces enough normal alpha-1 to protect the lungs. A full blown Alpha is known as PiZZ as they do not produce enough alpha-1 and it is also nonconforming which can cause problems as some of it stays in the liver. The other two more common types are PiMZ & PiMS for reduced levels of alpha-1 which are also non conforming but not to the same degree. There are many other defective proteins, but S and Z are the most common.

You may, therefore, have been told that your child has the phenotype PiMS, PiSS, PiMZ, PiSZ or PiZZ.
How Does This Affect Your Child?

New-born babies can often show signs that their livers are not functioning properly through the below signs:-

- Poor weight gain
- Jaundice
- Pale stools
- Dark urine

However, if it is known that the baby is a possible Alpha and these signs persist, specialists in liver diseases will need to investigate further to help in this complex area.

They will measure the level of alpha-1 antitrypsin (AAT) in the baby’s blood and other chemicals related to liver function.

From this they will choose the most appropriate treatment and also make an estimate of the likely progression of the condition.

Remember, jaundice is not uncommon in new-born babies so do not take this sign (or any other) in isolation as indicating liver problems. The type of jaundice related to the liver problem is different and can be identified by a simple blood test. But if in doubt always consult your Doctor.
Caring for Your Child
As your child grows you will want them to have the best opportunities to cope with, and overcome, possible complications stemming from alpha-1 antitrypsin deficiency.

If your child has liver problems or has received a transplanted liver then your medical specialist will have given you instructions on signs to look for and advice on how to respond when you see them.

As a parent you make decisions on your child’s behalf just as an adult Alpha should make for themselves. Amongst other things this means keeping your child away from airborne contaminants that can damage their lungs like smoke.

(A comprehensive list can be seen in our booklet “Newly Diagnosed”)

However, it is important not to be overprotective of your child. A child brought up to think of themselves as ‘sickly’ or ‘out of the ordinary’ is starting out at a disadvantage. Alpha children can live long and relatively healthy lives provided that they take sensible precautions. A positive attitude is very important.

Life is meant to be enjoyed not endured, so ensure your child enjoys life.

As your child grows up they can be slowly introduced to the do’s and don’ts of life as an Alpha. Start early with the habit of eating and enjoying a nutritious diet which is a gift that will last a lifetime. Pass on the knowledge that smoking is very dangerous for Alphas and this extends to passive smoking and vaping. An understanding of this may help your child resist peer pressures in their teenage years.
The Liver

The liver is the largest gland in the body and the only organ that can **repair and re-grow itself**.

It performs many functions from cleansing the blood of poisons to manufacturing bile to assisting in the digestion of fat in our food.

It helps breakdown carbohydrates in our food into a form that allows a quick release of energy when needed. This is why people with liver damage often lack energy.

The liver has many functions relating to keeping the blood clean and healthy. The one that concerns us is producing alpha-1 antitrypsin which protects the lungs from the activity of the enzymes released from white blood cells when the body is attacked by infecting agents such as germs, viruses and chemical irritants.

It is generally thought that the liver disease associated with alpha-1 antitrypsin deficiency is not caused by the alpha-1 deficiency in the bloodstream, but by excessive amounts of alpha-1 stuck in the liver cells.

Liver damage occurs in about 10% of infants born with the severe form of alpha-1. Some alpha-1 patients have cirrhosis of the liver. This affects very young alpha-1 children, as well as 12-15% of adult alpha-1 patients.
The Lungs:

Air enters the lungs as we breathe via the bronchi. The bronchi then divide into smaller and smaller tubes/branches (bronchioles), until finally becoming microscopic.

That air we breathe contains many unwanted particles, most far too minute to be seen with the naked eye. As adults, most of those particles/contaminates (called pathogens) are filtered out and discharged in mucus via the nose or mouth.

Some minute pathogens still get through and enter our lungs where they are dealt with by neutrophils (a type of white blood cell). The neutrophils devour the pathogens. Alpha-1 antitrypsin then “sweeps” up the neutrophils leaving the lungs clean and safe.

A lack of alpha-1 antitrypsin leaves those neutrophils free to attack other cells where they break down the elastin, (a protective coating on the lining of our lungs), degrading the elasticity of the lungs.

This can result in breathing disorders, such as Chronic Obstructive Pulmonary Disorder (COPD), in adults.

Cigarette, BBQ, open fire smoke, vehicle exhaust fumes and aerosol sprays are amongst the contaminants that we breathe in daily and that can damage our lungs if not fully protected.
Care of the Liver:

At this time there is no single treatment that can be prescribed for liver damage in children caused by AATD. Although in mild cases of liver distress treatment may be available to relieve the side effects.

Drugs may also be used to correct imbalances in the levels of substances processed or created by the liver. This is a complex area and each child is different and so the hepatologist (or liver specialist) will make an individual judgement.

As the liver processes many of the constituents of the food that we eat it is essential to eat a healthy and balanced diet, as well as follow all dietary advice from your hepatologist or specialist dietician if you have one. The general aim is to make it easier for the liver while providing sufficient nutriments to help the body thrive as the child grows.

N.B.

*There may be other conditions or intolerances that must be taken into account when planning a diet. These are general guidelines and your clinician/dietitian will be able to advise you on this.*
Care of the Lungs:

In the very early years your babies lungs are extremely vulnerable. Therefore, it is vital that you ensure they avoid breathing any unnecessary airborne contaminates such as cigarette smoke. This will stand them in good stead and possibly avoid the onset of COPD in later years.

As your child grows you will need to gradually introduce good habits into their lives, such as avoidance of any airborne contamination. This is vital for their future wellbeing.

Their lungs will remain vulnerable for the rest of their lives. So good habits need to stay with them.
Transplants:

Only in extreme cases of liver failure is there an option of a transplant. A “living donor transplant” is a possibility where a suitable healthy adult is prepared to donate a part of their liver. This is an increasingly popular option for liver transplantation in children. Experience has shown that the risks to both the donor and patient are minimal for what is a relatively simple surgical procedure. However, since AATD is a genetic condition some close family members may also be Alphas and therefore will not be able to be donors.

Lung transplants usually occur later in life when the lungs struggle to provide sufficient oxygen to supply the body.

Lung transplants are not as simple as a liver transplant and require the transplant of the whole lung. A difficult and complex operation that is a last resort to give relief especially where COPD is concerned. It is often difficult to find a match and can take several years before a successful one is found, so patience may be required. That being said lung transplants are usually very successful and can offer an excellent quality of life.
. . . and Care for Yourself

Your child is an Alpha, therefore so are you and/or your partner. If you have not already been tested then ask your doctor to perform the test.

Alpha-1 affects everyone differently and some Alphas live their lives and have no symptoms of liver or lung diseases. This may be due to chance but all Alphas should follow a few simple guidelines to help chance along:

- **Keep your body healthy** with a balanced diet and regular exercise. Infections and other illnesses can be more challenging to Alphas than to most people. So improve your defences.

- **Smoking** is especially harmful for people with AATD. Tobacco smoke releases increased amounts of a lung-damaging enzyme that destroys more lung tissue and what little AAT may be present in the lungs. **If you smoke, you should quit as soon as possible.**

- Avoid open fires, petrol fumes, paints, solvents, dust, etc. Heed any air quality warnings, such as high pollen counts, especially in the hot summer months.

- Try to avoid contact with anyone who has a cold or flu.

- Ask your GP if they recommend a yearly flu vaccination.

- Some GPs and Alpha Specialists recommend having hepatitis A & B vaccine to help protect the liver. It might be an idea to ask your doctor about this.
For more information please visit:
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