

NOT The Only Alpha-1

**A Teenager's
Guide to Alpha-1
Antitrypsin Deficiency**



Acknowledgements:

The Alpha-1 Association acknowledges the following individuals for their support in the development of this publication:

Brittany McLarney, MS – Author

Jeff Byrd – Illustrator

Reviewing Committee:

Mark Brantly, M.D., Marlene Erven, Cathey Horsak, Michael Krowka, M.D., Dawn McGee, MS, CGC, Genevieve Paul, Charlie Strange, M.D., Don Rabush, Ed.D, Ron Sokol, M.D. and Jeffrey Teckman, M.D.

Introduction

This book is for adolescents who have Alpha-1 or know someone who does. There's a lot of information in this book and it would be a good idea to write down any questions you may come up with while reading. There is a page in the back of the book with space to write down your questions so you can remember to ask your parents or doctor.

Also, at the end of this book is a list of all the words in **bold** so you can look up their meaning quickly if you happen to forget.

The last important page to mention at the end of the book, is a list of different resources you can use to meet other Alphas or get your questions answered.

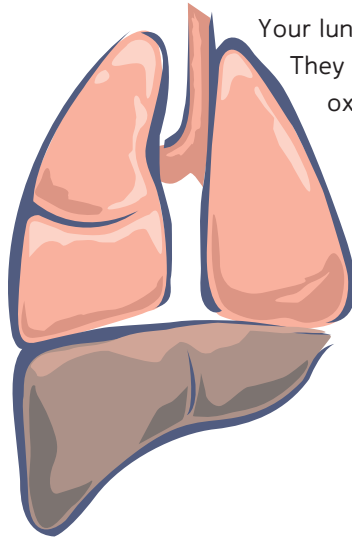
What is Alpha-1 Antitrypsin Deficiency?

Alpha-1 Antitrypsin Deficiency (Alpha-1) is a condition people are born with. Alpha-1 can cause liver disease in infants, children, or adults and lung disease in adults.



How do The Liver and Lungs Work?

Your liver plays many roles. First, your liver works as a filter. When blood flows through your liver, it takes out anything bad that is in the blood and gets rid of it. Your liver also plays an important role in digestion as well as storing important substances your body needs. These are just some of the important roles your liver performs for your body.

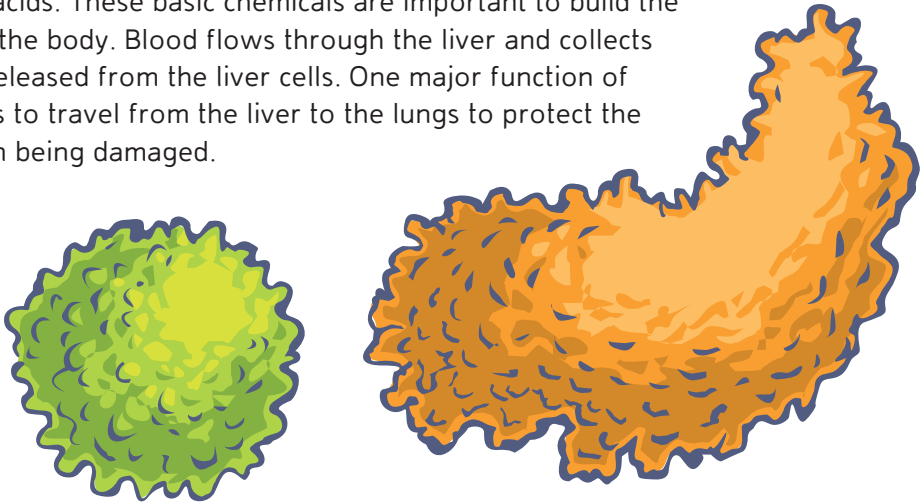


Your lungs exchange some gases in your body.

They get rid of carbon dioxide and bring in oxygen. When you breathe in, your lungs fill with air and when you breathe out, you exhale the air rich in carbon dioxide. Your blood vessels in your lungs and the heart distribute the oxygen throughout your whole body.

What is different in the liver and lungs in people with Alpha-1?

Alpha-1 Antitrypsin is a **protein that we will also call Alpha-1**. A protein is a type of molecule in the body made up of amino acids. These basic chemicals are important to build the tissues of the body. Blood flows through the liver and collects proteins released from the liver cells. One major function of Alpha-1 is to travel from the liver to the lungs to protect the lungs from being damaged.





In people with Alpha-1, the Alpha-1 protein is the wrong shape and gets stuck in the liver which can cause liver problems. Since the protein is stuck in the liver, it cannot get to the lungs to protect them. This can cause problems with the lungs.

Some of the symptoms associated with the liver disease can include **jaundice**, swelling, poor growth, and feeling weak or tired. Jaundice is when the eyes and skin of a person have a slight yellow color.

Lung disease symptoms typically occur in adults. These symptoms include, shortness of breath, coughing, **bronchiectasis**, and frequent chest colds. Bronchiectasis is an illness that causes the tubes leading to the lungs to be permanently widened and leads to infections.

Just because a person has Alpha-1 does not mean they will definitely have liver or lung problems. Alpha-1 increases the risk to develop problems with the liver and lungs so it is important to understand the symptoms, and it is just as important to know what things you can do to try to keep your liver and lungs healthy.

Alpha-1 runs in families

Alpha-1 is an inherited condition. This means Alpha-1 is passed down in families. There are many different conditions that are passed from parents to their children, and Alpha-1 is one of these conditions.

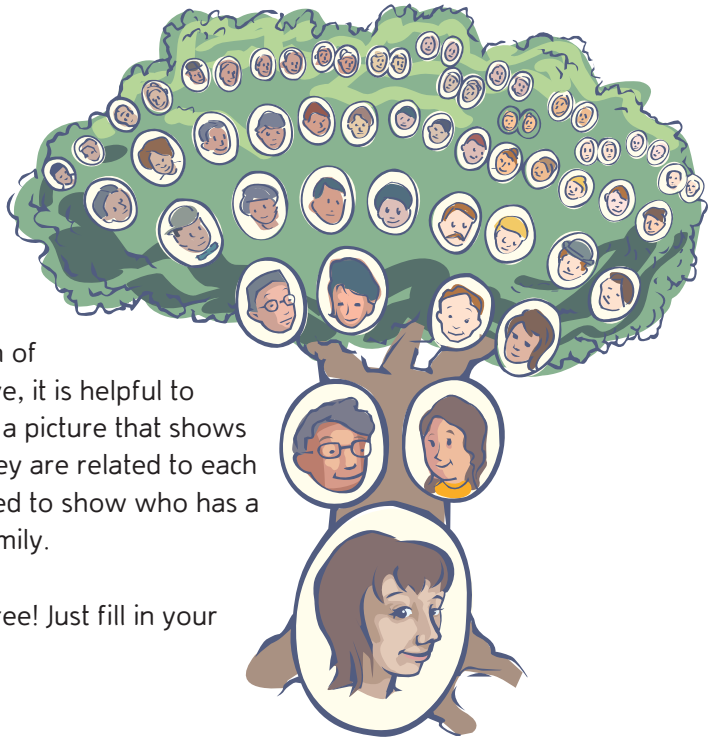
The way conditions are passed down in families is through **genes**. Genes are found in every cell of the body and tell the body how to work. Genes come in pairs because you get one copy from your mom and one from your dad. Genes determine everything about you, including how tall you are, your eye color, the shape of your ear, and if you get some conditions.





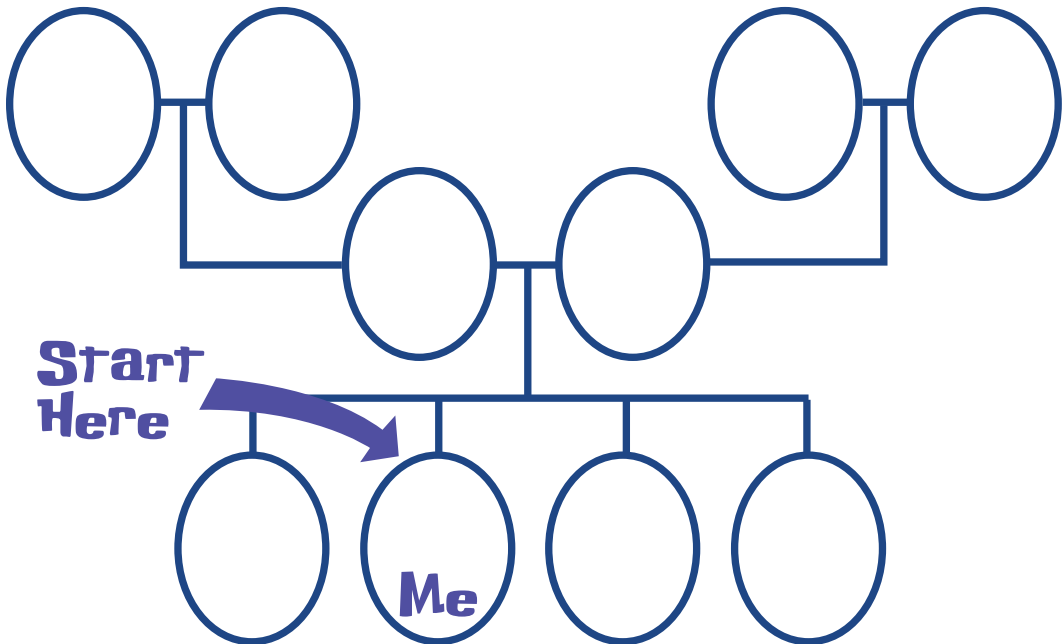
A change in a gene is called a **mutation**. People with Alpha-1 have mutations in their Alpha-1 gene that stops the gene from working correctly. Some mutations cause a gene not to work properly and other mutations do not cause any problems. The different forms of a gene are called alleles.

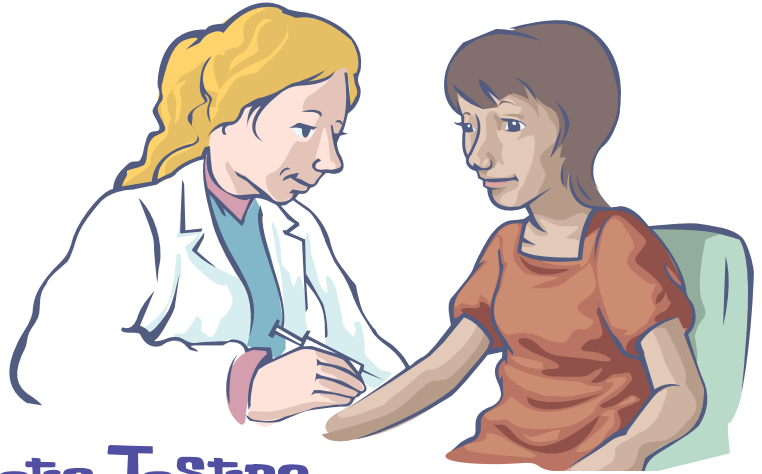
There are over 100 different **alleles** for the Alpha-1 gene. However, the three most common alleles are M, Z, and S. M is the normal allele for the gene and Z and S are the two most common alleles that cause the deficiency. Remember each gene comes in a pair, one from mom and one from dad. If both parents pass on an M, the child will be considered MM (two M alleles of the gene) and will not have Alpha-1. If one parent passes on an M and the other a Z, the child is MZ and is a **carrier**. A carrier is a person who has one normal allele and one allele with a mutation. A carrier for Alpha-1 typically does not get sick unless they are exposed to harmful irritants. These irritants will be discussed in more detail later in this book. The other type of carrier for Alpha-1 is MS. When someone is MS, they inherited the M allele from one parent and the S allele from the other parent. If someone inherits two copies of a mutation in their Alpha-1 genes, SZ or ZZ, then they have Alpha-1. This means they inherited a copy of the mutation from each parent.



To understand what combination of the gene for Alpha-1 you may have, it is helpful to draw a family tree. A family tree is a picture that shows everyone in the family and how they are related to each other. A family tree can also be used to show who has a condition and how it runs in the family.

You can draw your own family tree! Just fill in your family member's names.

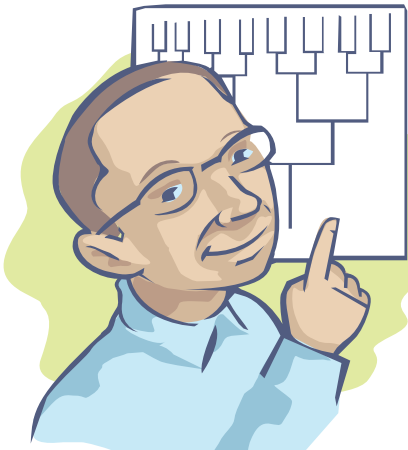




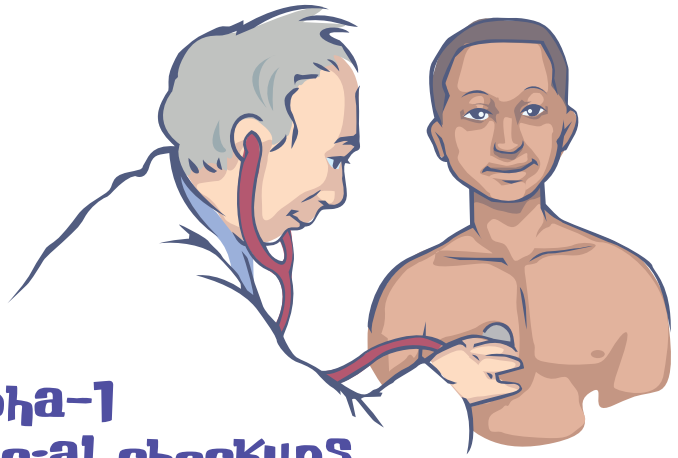
Genetic Testing and Genetic Counselors

How is Alpha-1 diagnosed?

Alpha-1 can be suspected based on your health, but the only way to know for sure is through a blood test. The blood test can be done from a **blood draw** or a **finger prick**. A blood draw uses a needle to take blood from your arm, while a finger prick uses a small needle to poke your finger so a few drops of blood can be collected. The sample of blood is then sent to a lab that can tell if you have Alpha-1 or not.



Before getting a genetic test your family may choose to talk to a **genetic counselor**. A genetic counselor is a person who understands how genes are passed down in families and can help your family determine if genetic testing is right for you and your family members. A genetic counselor will talk with you and your family to see if Alpha-1 is in your family. The genetic counselor will even draw out your family tree to see how Alpha-1 is being passed down in the family.



People with Alpha-1 need to get special checkups.

A **Liver Function Test (LFT)** may be done to check for liver damage. This test involves blood work that will let doctors look at certain proteins that may be affected by a damaged liver.

Another test may involve ultrasound. An **ultrasound** is a special, non-painful way to take pictures of your internal organs. Individuals with Alpha-1 and symptoms may need to get ultrasounds done to check their liver.

The last liver test we will talk about is a **liver biopsy**. A liver biopsy involves being given medication to numb your skin and make sure you do not feel any pain. Once you cannot feel any pain, a needle is put through your skin to the liver to take out a small piece of the liver. This piece can then be looked at by a doctor to see if the liver is normal or not.

The lung test a doctor might perform on you is called a **Pulmonary Function Test (PFT)**. A PFT measures how well the lungs are working by having you breathe into a mouthpiece that is connected to a special machine. This test looks at how much air the lungs take in and how much air is exhaled. It also looks at how well oxygen is passed around the body.

Irritants that should be avoided

Since Alpha-1 can cause lung and liver disease there are some irritants that you should avoid. Irritants are things that bother people; for example, pollen is an irritant that can cause people to sneeze and be itchy. For most people irritants do not cause permanent damage, but in Alpha-1, these irritants can be serious. Some irritants that should be avoided are:

- Cigarette smoke
- Dust and dirt

It is less clear that the following cause lung injury but can cause symptoms:

- Fumes
- Powders
- Mold and mildew
- Household cleaners
- Bug sprays
- Fireplace smoke
- **Aerosolized products**– These are products that are sprayed out of a can like hairspray, air fresheners, or perfume.

In order to avoid these irritants there may need to be some lifestyle changes. The goal is to use products that are safer and

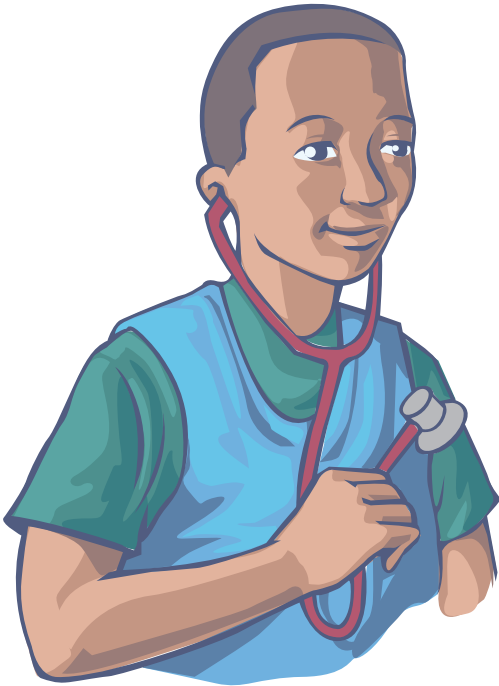
to wear a mask when these irritants can not be completely avoided. An example is that patients with Alpha-1 should never smoke cigarettes and should avoid being exposed to secondhand smoke.



Keeping Your Liver Healthy

Avoiding these irritants is the first step to staying healthy, but it is also important to start good habits in order to have a healthy future.

Eating a well-balanced diet can help you stay healthier. Just as important as eating correctly is exercising. Exercising is important for everyone, but it will help people with Alpha-1 breathe easier. Start with light, easy exercises and work up to harder, more intense exercise. If you are not sure what you should be eating or what exercises to do, ask your parents and doctor for help. Being overweight can cause fat to build up in the liver which may lead to liver scarring.



Remember, you should always follow the directions and recommendations of your doctor. It is necessary to take all medications and get all vaccines so you can stay as healthy as possible. Hepatitis viruses (Hepatitis A and B) can infect and damage your liver but can be prevented by vaccination. The vaccines will help you avoid infections and illnesses that could make you sick. Hepatitis viruses may also be transmitted through needles so it is important to not get tattoos or piercings from anyone but a professional who uses sterile needles.

Drinking alcohol can damage anyone's liver so it is smart not to drink beverages containing alcohol (beer, wine, hard liquor). Some herbs that you may be able to buy in health food stores may also damage the liver – you should check on this before taking them.

Another important thing for you to think about is your future. You want to avoid careers that have any of the irritants we listed. You are still young but you are going to want to start being healthy now and continue to be healthy in your future.



Common Symptoms to look for with Alpha-1

There are many different symptoms that can go along with Alpha-1. Each person with Alpha-1 may have different symptoms and some people won't get sick at all. Some of the most common symptoms are:

- Trouble breathing
- Wheezing
- Coughing a lot
- Recurrent chest colds
- Not being able to exercise for long amounts of time
- Year-round allergies
- Unexplained liver disease
- **Jaundice**– This is when the eyes and skin have a slight yellow color.
- **Bronchiectasis**– This is an illness that causes the tubes leading to the lungs to be permanently widened. This can lead to infections.
- Coughing up blood

It is important to tell your parents and your doctor if you are experiencing any of these symptoms. It is important for your doctor to know what symptoms you have so they can know if you are getting sick. Also, if your doctor knows what is bothering you, he or she can help you feel better.

Treatments and Surgeries for Alpha-1

One of the most common treatments for Alpha-1 is **augmentation therapy**. This treatment uses a needle to place the alpha-1 protein directly back into the body. This helps bring the levels of the protein in the blood back up which can help the lungs function better. This treatment does not help with the liver. Augmentation therapy is done typically once a week. This treatment is for individuals that have Alpha-1 and have lung disease. Therefore, this medicine is not given to children since the lung disease is not typically present in children.

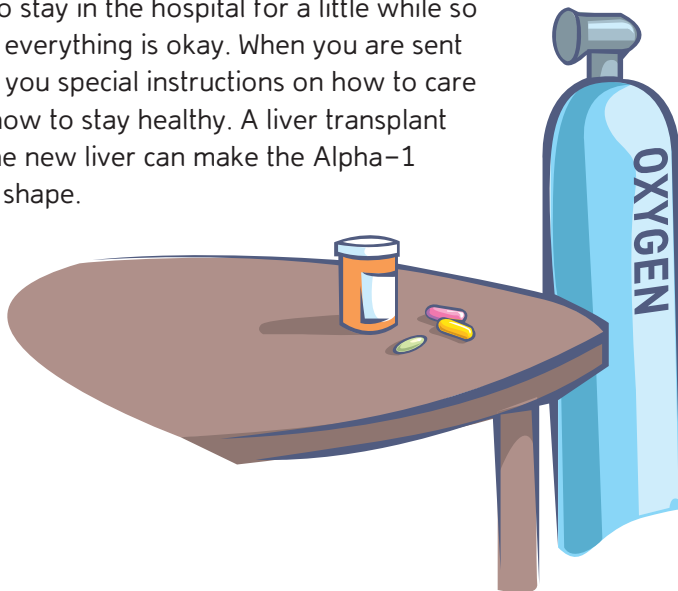
Other common treatments are corticosteroids and antibiotics. Corticosteroids are used to minimize chest symptoms while antibiotics are used to stop infections. Infections can speed up the damaging effects to the liver and the lungs, so it is important to avoid these problems.

If you are having difficulty breathing, your doctor may prescribe medication from an inhaler. An inhaler is a container that can release medicine as a gas that you can breathe in.

Some people with Alpha-1 may even need an oxygen tank. An oxygen tank is a container that delivers extra oxygen through a tube in the nose. This is not painful, but it does help a person to

breathe easier. An oxygen tank is used when the lungs become very damaged.

When the damage to the lungs or liver becomes too serious, a **transplant** may be necessary. A transplant is when a sick organ, like a liver or lung, is removed and replaced with a healthy organ. This requires an operation and you are not awake. After a transplant you will need to stay in the hospital for a little while so the doctor can make sure everything is okay. When you are sent home the doctor will give you special instructions on how to care for your new organ and how to stay healthy. A liver transplant cures Alpha-1 because the new liver can make the Alpha-1 protein that is the correct shape.



How to feel about Alpha-1 and who should know

You may feel different about Alpha-1 every day, and that is normal. There are no right or wrong ways to feel about this diagnosis. Some days you may not even think about Alpha-1 and other days you may feel nervous. The best thing to do if you are feeling upset or nervous about getting sick, is to talk to an adult you trust. You should not feel embarrassed about how you are feeling because chances are other people with Alpha-1 feel the same way.

It is also up to you to decide who you want to know about your diagnosis and when they find out. Of course your parents and doctor will know, but you can tell your friends when you are ready to. If you decide you do want to tell your friends about Alpha-1, it is a good idea to talk to a trusted adult first to plan out what you will say. It can feel strange teaching your friends about a condition they have never heard of, but once you explain it to a few people it will get easier.

Glossary

Aerosolized products– products that are sprayed out of a can like hairspray, air fresheners, or perfume

Allele– an alternative form of a gene. There can be multiple alleles for one gene, meaning there are multiple variations of the same gene.

Alpha–1 antitrypsin– a protein made in the liver that protects the lungs from being damaged

Augmentation therapy– this treatment uses a needle to put the alpha–1 protein directly back into the body. This helps bring the levels of the protein back up in the blood. This treatment does not help with the liver, but it is useful for the lungs. Augmentation therapy is typically done once a week.

Blood draw– a needle takes blood from a person’s arm

Bronchiectasis– an illness that causes the tubes leading to the lungs to be permanently widened. This can lead to infections.

Carrier– a person who has one normal allele for a gene and one allele with a mutation

Glossary (continued)

Finger prick– a small needle pokes a finger so drops of blood can be collected

Genes– found in every cell of the body and tell the body how to work. Genes come in pairs because you get one copy from your mom and one from your dad.

Genetic counselor– a person who understands how genes are passed down in families and can help your family determine if genetic testing is right for you and your family members.

Genetic test– a test that takes blood from a person by a blood draw in the arm or a finger stick and looks at their genes

Jaundice– when the eyes and skin have a slight yellow color.

Liver biopsy– a test where a needle is put through the skin to the liver to take out a small piece of the liver. This piece can then be looked at by a doctor to see if the liver is normal or not.

Liver Function Test (LFT)– a test to check for liver damage. This test involves blood work that will let doctors look at certain proteins that may be affected by a damaged liver.

Mutation– a change in a gene

Protein– a type of molecule in the body that helps make all the different parts like skin, muscle, and many more

Pulmonary Function Test (PFT)– a breathing test

Transplant– when a sick organ, like a liver or lung, is removed and replaced with a healthy organ

Ultrasound– a non–painful way to take pictures of organs inside a person’s body using soundwave

White blood cells– special blood cells that important in keeping you healthy

Resources

You may not know any one outside of your family who has Alpha-1, but there are a lot of other people with the same condition. There are some great organizations that can help you feel better about having Alpha-1 and can help you understand the condition even better. Here is a list of some useful organizations.

Alpha-1 Association

(800) 521-3025

www.alpha1.org

Alpha-1 Kids

(877) 346-3212

www.alpha1kids.org

Alpha-1 Association Genetic Counseling Program

(800) 785-3177

www.alpha1.org/support/

Alpha-1 Foundation

(877) 228-7321

www.alpha-1foundation.org/



Alpha-1 Kids

The Alpha-1 Kids program is dedicated to providing support, education and information for parents and families of children affected by Alpha-1.

For more information about the Alpha-1 Kids program, please contact us at:
Telephone: (877) 346-3212 • E-mail: info@alpha1.org



Toll-free: (877) 228-7321 • Local: (305) 567-9888
www.alpha1.org • E-mail: info@alpha1.org
3300 Ponce de Leon Blvd • Coral Gables, FL 33134