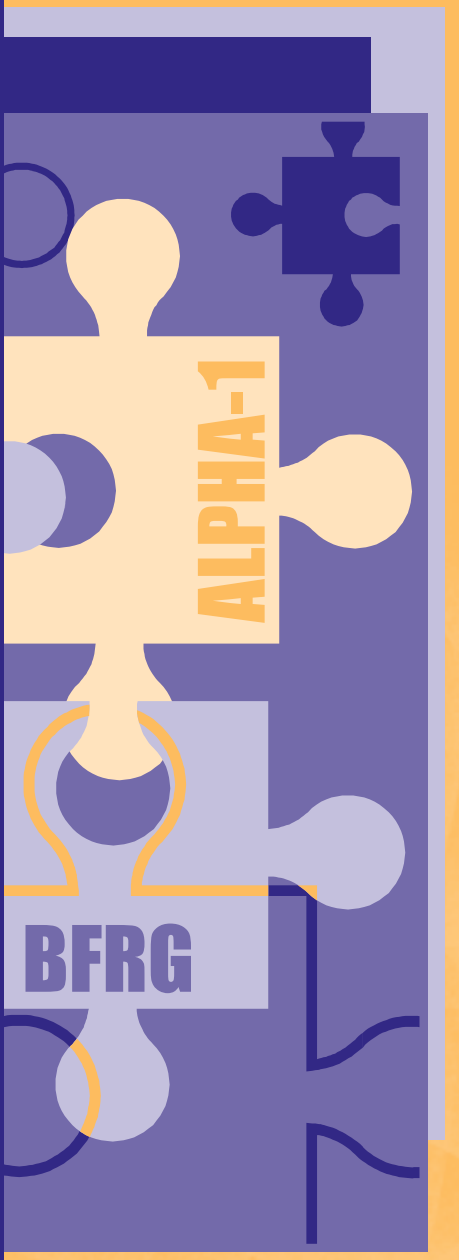
ALPHA-1 ANTITRYPSIN DEFICIENCY
A GUIDE FOR THE RECENTLY DIAGNOSED INDIVIDUAL

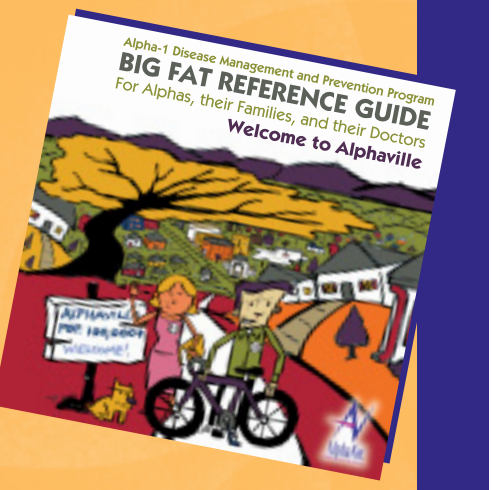
FORGING PARTNERSHIPS FOR A CURE

DO YOU NEED ACCESS TO INFORMATION, RESOURCES AND PEACE OF MIND?

THIS GUIDE IS YOUR KEY TO ALPHA-1



Do you understand Alpha-1 Antitrypsin Deficiency? Have you labored over a medical dictionary or struggled with the terms that doctors use? Or wondered what you can do to stay healthy? **The Big Fat Reference Guide (BFRG)** will help you unlock these answers. It's the most comprehensive resource for living with Alpha-1 available anywhere.



With the **BFRG**, you can understand, manage, and live with Alpha-1. It has detailed information on genetics and on understanding lung disease, discussions on environmental risk factors, suggestions on diet, nutrition, activity and fitness, and much, much more. Plus there is practical information about insurance and disability, and explanations about key terms and the various diagnostic tests.

This invaluable resource, available on-line at no cost, was developed by AlphaNet, a not-for-profit health management organization dedicated to improving the lives of patients with Alpha-1. Written by patients and healthcare professionals, it is part of AlphaNet's Disease Management and Prevention Program (ADMAPP), a key service staffed by Alphas to serve Alphas.

Use the **Big Fat Reference Guide**, written in language that you can understand, to learn more and share the insight with your family.

Register at ***www.alphanet.org*** to access the **BFRG** today.

This ad courtesy of

ALPHA-1 FOUNDATION

FORGING PARTNERSHIPS FOR A CURE

The Alpha-1 Foundation acknowledges the significant contribution of AlphaNet in developing this comprehensive program for Alphas, and for providing over $18 million to support our research mission.

THE DIAGNOSIS OF ALPHA-1

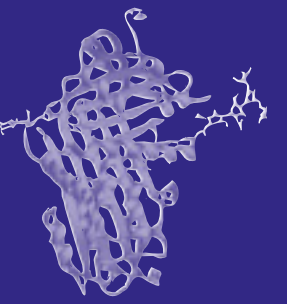
Alpha-1 Antitrypsin Deficiency, also known as AAT Deficiency or Alpha-1, is a medical diagnosis that should lead to open discussions with your doctor and family. Many Alphas, as individuals diagnosed with Alpha-1 are known, live full and productive lives. The following information is designed to help you learn about this inherited disorder so that you and your family can take the necessary preventive measures to stay as healthy as possible.

Share this information with your family and your healthcare professional. You may also want to seek professional genetic counseling and contact organizations listed at the end of this brochure to obtain more information.

UNDERSTANDING ALPHA-1

Alpha-1 is an inherited disorder that it is present at birth. Alpha-1 may result in the development of serious lung and/or liver disease. Approximately 100,000 people in the United States are estimated to have Alpha-1.

In typical individuals, large amounts of the alpha-1 antitrypsin (AAT) protein are made in the liver and released into the blood. The proteins are made from normal genes, which are an inherited component of every cell that direct specific biological functions. In affected individuals, altered genes make abnormally-shaped AAT proteins. The abnormal protein is retained in the liver and cannot be fully released into the blood, creating a deficiency in your body.



Structural representation of alpha-1 antitrypsin

ALPHA-1 IS:

- A genetic disorder inherited from one's parents.
- Indicative of a reduced amount or lack of the AAT protein in the blood.
- A cause of chronic obstructive pulmonary disease, even in people who have never smoked.
- A leading cause of genetic liver disease in infants and children.
- Often misdiagnosed.

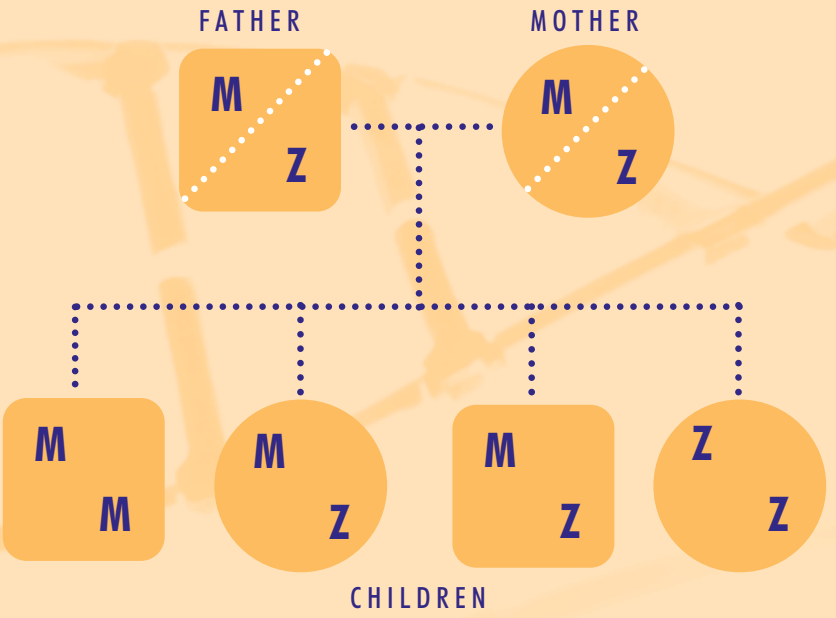
ALPHA-1 CAN CAUSE:

- Lung disorders, including asthma-like symptoms, chronic bronchitis, emphysema, or bronchiectasis. These problems, usually seen in adults, are caused by the AAT protein deficiency and are commonly grouped under the term COPD, which means chronic obstructive pulmonary disease. Normal AAT protein levels protect the lung tissue from the destructive effects of white blood cells circulating in that organ. When levels of the protein are low or absent, lung deterioration can occur.
- Liver disorders, including elevated liver enzymes, neonatal hepatitis, chronic liver disease, cirrhosis, and liver cancer. These problems are due to the accumulation of the abnormal protein in the liver cells. Although less common, Alpha-1 related liver disease can be seen at all ages beginning in infancy. Liver cancer is usually seen only in adults.
- Panniculitis, a skin disease created by an excess of white blood cell products. Panniculitis frequently causes painful lumps under or on the surface of the skin.

GENETICS OF ALPHA-1

In each of us, the normal AAT protein is made from a pair of genes obtained from one's parents. Each parent donates one alpha-1 gene. The normal protein is made from the M gene. [See Testing for Alpha-1.] For unknown reasons, there are close to 100 altered or abnormal variants of the M gene, also called alleles, but only a few can cause serious lung, liver or skin disorders. Two important alleles are the S and Z gene variants. An individual with different gene variant pairs, such as the MZ genes, is called a heterozygote. Someone with similar gene variant pairs, such as the ZZ combination, is called a homozygote.

Individuals with one normal and one variant alpha-1 gene are called carriers. Alphas can have parents who are both carriers, one parent who is a carrier and one who is severely deficient, or two parents who are severely deficient. Since half of an individual's genes are inherited from each parent, the diagram below shows four possible genetic combinations for children of two Alpha-1 carriers, meaning each parent has one normal M and one altered gene, in this case the Z allele. It also illustrates how carriers who may be unaware of their risk might have a child with the most severe form of the disease, the ZZ combination.



These are the most common variants but there are many other possibilities. You may wish to read the Alpha-1 Foundation brochure titled, *"What Does It Mean to Be an Alpha-1 Carrier"* and then discuss your specific test results with your healthcare professional.

RISKS ASSOCIATED WITH COMMON GENETIC VARIANTS

NORMAL (MM):
Does not have the disorder and does not carry any altered alpha-1 genes.

CARRIER (MS):
It is unclear whether there is a risk for developing disease symptoms but this individual does have an altered alpha-1 gene. Most studies do not indicate an increased risk for disease.

CARRIER (MZ):
An MZ carrier may have a mild AAT protein deficiency but rarely develops disease symptoms even though he/she does carry an altered gene.

ALPHA-1 (SZ) OR (ZZ) OR (NULL NULL):
Moderate (SZ) to severe (ZZ, Null Null) AAT deficiency. This individual could develop Alpha-1-related disease and has two altered alpha-1 genes.

TESTING FOR ALPHA-1

The diagnosis of Alpha-1 is usually determined by three rather sophisticated laboratory tests that require a small sample of blood. The tests:

1. Measure the level of the AAT protein in your blood
2. Characterize the phenotype of the protein in the blood, and
3. Determine the patient's alpha-1 genotype.

ALPHA-1 BLOOD LEVELS

Most commercial medical laboratories can measure AAT protein blood levels. Clinically-significant deficiency exists when the blood levels are less than 80 milligrams per deciliter (mg/dl) or 11 micromolar (μM). Patients with such low levels are called Alphas or are frequently noted to have Alpha-1.

ALPHA-1 PHENOTYPE

This test characterizes the type of alpha-1 gene (for example, MM, MZ, SZ, or ZZ) that circulates in your blood by visualizing the movement of the protein in a special electric field. Close to 100 types of alpha-1 gene variants producing AAT protein have been identified, yet each person has only one type of protein. The SZ and ZZ phenotypes are the most common genetic patterns associated with the lowest blood levels (the most serious deficiencies) of the AAT protein.

ALPHA-1 GENOTYPE

The genotype is determined through a test that identifies the actual genetic variants producing the AAT protein. This test detects the S and Z alleles with great accuracy.

Most medical laboratories report AAT blood levels and phenotypes; special laboratories must determine genotypes. Your physician can suggest the best testing strategy for you and your family members.

LUNG SYMPTOMS OF AAT DEFICIENCY

If you have respiratory symptoms, you and your physician may observe:

- Shortness of breath while at rest or with exercise
- Wheezing, persistent cough
- Recurrent lung infections
- Persistent sputum (or phlegm) production
- History of suspected allergies and/or asthma
- Sinus infections

POSSIBLE LIVER SYMPTOMS

- Increased liver enzymes detected via a blood test ordered by a physician
- Jaundice or yellowing of the eyes and skin
- Enlarged liver and/or spleen, which may be noted by the patient, parent of child, or physician
- Ascites or fluid collection in the abdomen
- Cirrhosis, an excessive accumulation of scar tissue in the liver noted by a physician
- Vomiting of blood
- Persistent itching
- Noticeable change in energy level or becoming easily fatigued
- Blackish, purplish, dark or pale-colored stools
- Poor appetite

EVEN ALPHAS MAY NOT HAVE SIGNS OR SYMPTOMS

Even without symptoms, you should follow the recommendations in the next sections. If you practice a healthy lifestyle and obtain appropriate medical care, you may remain healthy. However, this does not mean that you will not have symptoms in the future, so monitoring of your medical condition by a healthcare provider is suggested. You may request a copy of the Alpha-1 Foundation's Healthcare Provider's Guide to give to your doctor.

WHAT DOES HAVING ALPHA-1 MEAN TO ME?

The following sections deal with issues that are potentially major concerns for Alphas. The resources listed at the back of this brochure provide advice and support as does AlphaNet's *Big Fat Reference Guide* (see inside front cover). In addition, genetic counseling can provide the support and knowledge needed for informed decisions.

HAVING ALPHA-1 MAY REQUIRE:

CHANGES IN LIFESTYLE

If you or your child has Alpha-1, it may be necessary to make lifestyle changes to stay healthy, including:

- Quitting smoking and keeping the Alpha adult or child away from second-hand smoke
- Avoiding exposure to dust and fumes
- Exercising regularly
- Eating well
- Drinking alcohol with caution, if at all. The consumption of alcoholic beverages can cause damage to the liver in normal people. Many authorities recommend low, infrequent or no alcohol consumption for ZZ patients, and patients with any indication of liver damage should avoid alcohol completely.
- Review labels of over-the-counter medications, vitamins or herbal supplements carefully. Avoid products with acetaminophen and alcohol, both of which can injure the liver.
- Advise your healthcare provider if you are taking vitamin supplements or herbal products.

ENVIRONMENTAL AWARENESS

Environmental recommendations include avoiding pollutants that irritate the lungs, as well as avoiding liver toxins. Individuals should assess their home and work environments carefully, and consult with an occupational medicine specialist if indicated. Examples of such environmental irritants are:

- Cigarette smoke, from personal smoking or second-hand tobacco smoke
- Industrial and occupational pollutants such as dust, flower and tree pollen, ash, volatile compounds, fumes and other allergens
- Air pollution
- Wood-burning stoves
- Fumes from cleaning solvents such as bleach, ammonia or household and industrial cleaners
- Paints and/or toxic agents
- Precautions should also be taken when handling chemicals and other materials, as those may be absorbed through the skin. The liver detoxifies poisonous chemicals that enter the body. If the liver is damaged, the detoxification process is altered.

INCREASED DOCTOR VISITS

Alphas should seek expert medical treatment and may need to visit their healthcare provider more often.

VARIOUS TREATMENTS

Alphas have various treatment options, depending upon their symptoms. The most common treatments are:

- Behavioral and lifestyle modification
- Medication therapy for lung problems
- Surgical therapy for lung disease
- Procedural treatments for the complications of liver disease
- Organ transplantation

WHAT DO I DO NOW?

Individuals with Alpha-1 should NEVER smoke. Evidence shows that smoking tobacco products significantly increases the risk and severity of emphysema in Alphas and may decrease their lifespan by as much as ten years or more. Exercise and nutritional programs also contribute to maintaining a healthier body. You must aim to achieve and maintain a healthy lifestyle by adopting the following recommendations:

SMOKING CESSATION

If you smoke, it is extremely important that you quit. This is necessary because smoking destroys the small amount of AAT protein in the lungs of those affected by the disorder and attracts white blood cells that have cell-damaging enzymes. These damaging enzymes speed the development of lung disease. If you are an Alpha, your lungs do not have the normal defenses against white blood cells. If your child has been diagnosed with Alpha-1, it is important to protect them from exposure to second-hand smoke. Educate your children on the dangers of smoking and the importance of avoiding second-hand smoke.

AVOID POLLUTANTS & INFECTION

You should avoid occupational and environmental pollutants that can be inhaled, including pollen, dust, or organic fumes, and second-hand tobacco smoke. These substances can irritate your lungs, and cause or worsen lung problems. Chemicals can also be absorbed through the skin and thus damage the liver. Avoid air pollution and aerosolized sprays at all times. It is also important to realize that you may encounter pollutants and infections, both at home and at work.

IN THE WORKPLACE

Avoid exposure to inorganic or organic dust, (coal, hay, etc.) or irritating gases (chlorine, isocyanates, etc.). Seek the healthiest possible work environment. Demand clean indoor air, with proper ventilation and filtration systems, and avoid second-hand tobacco smoke. Wear protective clothing such as rubber gloves when in contact with chemicals or other agents, many of which can be absorbed through the skin. Read labels closely. As previously suggested, consult with an occupational medicine specialist if you have any concerns about the specific effects of work-related exposures to your lungs and liver.

IN THE HOME

You should avoid:

- Household chemicals
- Respiratory irritants from wood-burning stoves, dust and pollen, or second-hand smoke
- Chlorine and ammonia, which are found in common household cleaning products
- Pesticides
- Pet dander

Since bacterial and viral infections are harmful to the lungs, avoid contact with sick or infectious people whenever possible. Hand washing with soap is the single most effective way to avoid both contracting and spreading infectious diseases. Carry a hand disinfectant gel with you for times when hand washing is not possible.

DEVELOP AN EXERCISE PROGRAM

Routine exercise improves mental outlook, stamina and physical well-being. Exercise is essential to all Alphas. It is important to exercise muscles in the chest and upper body that are related to breathing as well as the large muscles of the legs.

WALKING PROGRAMS

Walking programs (particularly in climate-controlled indoor shopping malls), strolling, swimming, and/or biking may improve your lung function and endurance.

A PULMONARY REHABILITATION EXERCISE PROGRAM

A Pulmonary Rehabilitation Exercise Program (PREP) is highly recommended for Alphas with all stages of lung disease or pulmonary problems. A PREP that includes exercise, breathing retraining, education, smoking cessation, and nutritional counseling may help you achieve your fullest level of activity. As with all exercise programs, a PREP should be discussed with and recommended by your healthcare professional.

PERSONAL EXERCISE PLAN

You may want to have a personally tailored exercise program that is carefully monitored by your healthcare provider and/or exercise specialist. Start exercising slowly and build the intensity of your program over time as your tolerance increases.

DEVELOP A NUTRITION PROGRAM

Proper eating habits may help to preserve lung and liver function; therefore you should establish or maintain good eating habits. Maintaining an ideal body weight, whether or not you have lung and/or liver disease, is important. Additionally, some scientific research indicates that people with lung disorders need to consume more calories than "lung-healthy" people. If you have lung and/or liver problems, consider working with a nutritionist or registered dietician to set up an individualized nutrition program.

The nutritional needs of patients with Alpha-1 related liver complications are highly individualized. Since fluid retention is common, sodium and protein intake may become a concern, and, good nutrition is advised. For instance, processed foods have high sodium content, so Alphas should read nutritional labels carefully. Vegetable sources of protein are better than those from animals. Vitamin A, B3 (Niacin), and iron supplements may stress an already compromised liver. In Alphas with signs of liver complications, fat absorption may be altered; therefore, your healthcare provider may recommend supplementing the diet with vitamins A, D, E and K. In the infant that is experiencing feeding difficulties, shows poor growth and a failure to thrive, a special formula is often recommended. Sometimes, total parenteral nutrition (TPN) may be necessary.

REDUCE STRESSORS:

Alphas report benefits with stress reduction techniques, including many relaxation exercises. These relaxation techniques may also contribute to a positive outlook on life and may prevent depression. Here are a few options to consider:

- Yoga
- Meditation
- Breathing exercises
- Muscle relaxation
- Biofeedback
- Visualization
- Hypnotherapy
- Positive thinking

WHAT ARE THE CURRENT TREATMENTS FOR ALPHA-1?

You may benefit from lifestyle modification. However, if you have lung and/or liver disease, seek expert medical care to treat your condition(s). There are specialized Clinical Resource Centers specializing in Alpha-1 throughout the United States. These centers are staffed with a healthcare team to provide medical treatment, and behavioral and lifestyle modification guidelines specifically for Alphas. To see if there is one in your area, please check the Foundation's website, www.alphaone.org.

VACCINATIONS:

- It is important for you to have annual flu shots. The use of these prophylactic or preventive vaccinations is of the utmost importance.
- The Pneumovax® vaccine may help prevent pneumonia. Consider repeating the pneumonia shot every six years.
- Discuss the vaccines for Hepatitis A and B with your healthcare provider.

AGGRESSIVE TREATMENT OF LUNG INFECTIONS:

It is important to notify your healthcare provider immediately when you suspect a lung infection. Because the lungs contain more white blood cells when you have an infection (and hence, more destructive enzymes), it may be necessary to take antibiotics to fight the infection.
These are some symptoms to watch for:

- Fever (with or without chills)
- Increased shortness of breath
- Increased coughing
- Changes in color or thickness of sputum (phlegm)

ADDITIONAL PREVENTIVE MEASURES:

Wash your hands frequently with soap to prevent the passage of viral or bacterial infections.

THERAPIES FOR ALPHA-1 LUNG DISEASE

ANTIBIOTICS

Bacterial infections in the lung can lead to a dramatic influx of white blood cells into the organ's tissue and airways, which may be one of the major causes of lung destruction in Alpha-1. To minimize this risk, many physicians advocate aggressive antibiotic treatment at the first signs of a lung infection. Even though 'pulmonary exacerbations' may not be caused by bacterial infection or may be due to a virus, which would not be expected to benefit from antibiotics, the benefits of this aggressive approach may, in some patients, outweigh the risks of antibiotic overuse. These risks include encouraging the growth of antibiotic-resistant bacteria, overgrowth of yeast and other agents that can lead to disease, and allergic reactions.

BRONCHODILATORS

Some of the symptoms of Alpha-1 are similar to common lung diseases such as asthma and COPD. Medications called bronchodilators, usually administered via inhalers, may be useful in relieving lung symptoms. Sometimes, different types of bronchodilators are combined to achieve maximum benefit. These medications allow better airflow in and out of the lungs by relaxing smooth muscle that surround the airways.

CORTICOSTEROIDS

Based on your healthcare provider's recommendation, the use of corticosteroids (or simply, steroids) can be an appropriate treatment for lung symptoms in some individuals. Steroids help reduce inflammation within and around the airways, and can be administered by inhalation, in pill form, or intravenously (into a vein). Steroids administered by mouth or vein are usually reserved to treat severe lung problems.

SUPPLEMENTAL OXYGEN

Supplemental oxygen can be important and life-saving therapy for individuals with low blood oxygen levels. Some people, however, need supplemental oxygen primarily during exercise or with sleep. For some, it is especially important when traveling by air or at high altitudes. Ask your healthcare provider about your need for this treatment.

AUGMENTATION THERAPY

Augmentation therapy, the process of receiving ATT protein that has been purified from the blood of human donors with normal alpha-1 genes, is appropriate for many Alphas with lung problems. As its name suggests, it increases, or augments, the protein levels in the blood, and may help to slow the loss of lung function. Augmentation therapy is usually given intravenously once per week.

Augmentation therapy is not a cure; it will not reverse lung damage that has already occurred, nor treat or prevent liver problems related to Alpha-1. It may help prevent lung problems from getting worse. Currently, augmentation therapy can only be prescribed for individuals with Alpha-1-related emphysema under supervised professional care.

SURGERY OPTIONS

Your healthcare provider may evaluate the need for one of the two types of surgery recommended for patients with severe Alpha-1: lung volume reduction and lung transplantation.

LUNG VOLUME REDUCTION

Lung volume reduction may improve breathing by physically removing some of the most damaged lung tissue. The benefit is variable and may last only a few years.

LUNG TRANSPLANTATION

Lung transplantation for one or both lungs is an option for some Alphas with severe lung disease. Lifelong drugs to suppress the immune system are required afterward.

As with all surgery, the outcomes and quality of life after such procedures depend on a number of issues specific to each person. Please consult with your healthcare professional about these options.

THERAPIES FOR ALPHA-1 LIVER DISEASE

AUGMENTATION THERAPY

There is no proven role for augmentation therapy to treat the liver manifestations of Alpha-1.

GENERAL TREATMENT OF LIVER COMPLICATIONS

It is important for parents, caregivers, or significant others to be aware and advised of any indication of possible complications related to liver disease. Additional discussion and references concerning the diagnosis and treatment of severe liver disease can be found in the brochure entitled *The Liver and Alpha-1*.

Once liver injury is identified in an Alpha, the first course of action is to evaluate your lifestyle for ingestion of potential liver toxins, such as alcohol, large doses of certain vitamins, and some medications. As with the pediatric population, careful follow-up of abnormal liver function is needed. Liver disease is treated symptomatically and preventatively. In adults, symptoms such as vomiting blood may occur suddenly with no prior indication of illness.

Substances in some medications may be harmful to your liver. These include both prescription and over-the-counter formulas with acetaminophen or alcohol, and nutritional supplements such as vitamins, herbs, and protein drinks. Make a list of any medicines you use and review it with your healthcare provider. Often, the liver injury can prove to be mild and temporary.

MEDICAL/SURGICAL PROCEDURES

There are several treatment options that may become necessary to improve the symptoms of advanced liver disease. These include:

- Large volume paracentesis (LVP), which is the removal of fluid from the abdomen.
- Banding or sclerotherapy of veins in the esophagus to reduce bleeding from distended or swollen veins. Banding involves using rubber bands to stop blood flow. Sclerotherapy involves the injection of a chemical irritant which causes blood flow to shift to nearby healthy blood vessels from the diseased vein.
- Portal vein decompression, which reduces the pressure in the blood vessels entering the liver from the digestive organs. This is a major surgery utilizing shunts to reroute blood flow to the liver and reduce the pressure in the blood vessel.

The latter two options help control the increased risk for bleeding in those with advanced liver disease.

LIVER TRANSPLANTATION

Liver transplantation can dramatically improve the symptoms of advanced liver disease caused by Alpha-1. Because the organ recipient will begin to produce the protein of the donor liver, alpha-1 protein levels should be normal after the transplant. The option of living-donor liver transplantation is available at some transplant facilities. If you decide to explore this option, check with the transplant center first to be certain that this choice is available to you.

OTHER ISSUES OF CONCERN FOR ALPHAS

Listed below are some issues that you may face after diagnosis. These are merely a starting point for discussion with your physician, genetic counselor, family or pastoral care advisor.

AWARENESS

Consider wearing a medical alert bracelet or necklace that lists important medical information.

PSYCHOSOCIAL/FAMILY SUPPORT

Q: WHAT DO I TELL FAMILY MEMBERS?

A: We recommend that you inform blood relatives of the test result because of the genetic nature of the disorder.

Q: SHOULD I URGE FAMILY MEMBERS TO BE TESTED?

A: After consulting with your healthcare provider, it is reasonable to encourage your blood relatives to seek testing. Because of the genetic nature of Alpha-1, your blood relatives could be carriers or have the disorder themselves. Issues related to genetic discrimination in employment and insurance need to be considered, however. Confidential medical testing for Alpha-1 is available through a research study at the Medical University of South Carolina (MUSC); call 1-877-886-2383 for more information.

HEALTH INSURANCE

Q: WILL THE ALPHA-1 DIAGNOSIS AFFECT MY HEALTH INSURANCE?

A: It may. The answer to this question depends on your current insurance coverage.

IF YOU ARE CURRENTLY INSURED, IT IS IMPORTANT THAT YOU EDUCATE YOURSELF ABOUT:

- Your specific insurance policy and benefits related to coverage and reimbursement
- Your lifetime maximum benefit, if any
- The laws of your state regarding mandatory coverage.

IF YOU ARE CURRENTLY UNINSURED: Alpha-1 may be considered a pre-existing condition, and insurance companies may not, in the future, be obligated to cover costs for this specific condition for some period of time. Consider getting professional advice and familiarize yourself with the insurance regulations in your state. Generally, you are obligated to inform an insurance company of any pre-existing condition when you apply for coverage.

EMPLOYMENT

Q: CAN I CONTINUE TO WORK?

A: The answer to this question usually depends upon two conditions:

- The present state of your health, and
- The possibility of unwanted airborne exposures such as dust and fumes at work, or other hazardous chemicals that might be in contact with your skin.

Work is good for your mental and emotional well-being. If, after discussion with your doctor, you are physically able to do so and can avoid occupational hazards, you should continue to work. Otherwise, your healthcare provider may suggest changing jobs to reduce these exposures. Please note that your eligibility to continue health insurance coverage may vary from state to state. If you change jobs after diagnosis, the issue of disclosure of your disorder may also affect your future coverage.

Q: WHAT IS THE ROLE OF DISABILITY INSURANCE?

A: If your physical condition does not allow you to work, discuss your eligibility for disability insurance payments with your healthcare provider and other professionals familiar with these benefits.

CONFIDENTIALITY

Q: WHO WILL KNOW THAT I HAVE ALPHA-1?

A: The results of your test will be included in your medical record. Although generally treated as confidential, insurance companies, healthcare facilities and other professionals may access this information, which may affect future employment and insurance benefits. You may obtain confidential Alpha-1 testing through the Medical University of South Carolina (MUSC).

Q: TO WHOM SHOULD (OR MUST) I DISCLOSE MY DIAGNOSIS?

A: You must make your own decisions about releasing this information. However, it is highly recommended that you tell blood relatives about their inherited risk of Alpha-1 and urge them to be tested. You should also inform future healthcare providers. You may be required to inform insurance companies if you change insurance policies.

Finding out about an Alpha-1 diagnosis can be an overwhelming and potentially upsetting experience. It is important to share this information with family and seek professional counseling, if necessary.

GLOSSARY OF TERMS

ALLELE	Another general name for the altered form of a gene. For example, the Z allele is an altered form of the M gene. There are many different alpha-1 gene alleles.
ALPHA-1 ANTITRYPSIN PROTEIN	The alpha-1 antitrypsin (AAT) protein is primarily made in the liver, which releases it into the blood-stream in typical individuals. The alpha-1 protein has many functions, one of which is to protect delicate tissue in the body from being destroyed by neutrophil elastase, a tissue-digesting enzyme most commonly found in circulating white blood cells. These enzymes are released into tissue when the white blood cells fight infection.
ALPHA-1 ANTITRYPSIN DEFICIENCY (ALPHA-1)	A genetic condition caused by the inability to pass the AAT protein out of the liver, which creates a deficiency throughout the body. People with Alpha-1 might develop liver problems or lung diseases such as emphysema, or a skin disorder known as panniculitis. Others do not have any symptoms or illness.
ASCITES	Fluid collection in the abdomen.
ANTIBIOTICS	Antibiotics are drugs that can kill or stop the growth of bacteria. Sometimes the term is used to describe drugs that can treat any infections such as those caused by bacteria, fungus, tuberculosis, and even viruses.
ASTHMA	A condition of the lungs characterized by widespread narrowing of the airways due to spasm of the smooth muscle, swelling of the mucous membrane lining the respiratory tract, and the presence of mucus in the inner spaces of the airway branches leading to the lungs.
AUGMENTATION THERAPY	Intravenous administration of the alpha-1 antitrypsin protein purified from human blood and given in sufficient amounts to protect the lungs from damage.
BILIRUBIN	Bilirubin is a by-product of red blood cell breakdown that is normally formed in the liver. It creates the yellow tinge of normal serum, the yellow-green hue of bile, the brown color in stools, and the yellow color of urine. When the liver is not functioning normally, the bilirubin level can rise, which causes jaundice, a yellowing of the eyes and skin.
BIOPSY	The term biopsy is used to describe both a procedure to remove tissue from an organ or a piece of tissue that is being examined under a microscope. There are three basic types of biopsies: a fine needle biopsy, a core needle biopsy, and a wedge biopsy.
BRONCHIECTASIS	Chronic dilation or widening of the bronchial tubes within the lung signals bronchiectasis. It is often caused by inflammatory diseases or obstruction and leads to chronic lung infection.
CHOLESTASIS	A backup of bile in the liver; may result in jaundice, dark urine, pale stools, and itching.
CHRONIC BRONCHITIS	A lung disease characterized by inability to move air in and out of the lung combined with the production of sputum on most days of the year. This is one of the diseases caused by cigarette smoking.
CHRONIC OBSTRUCTIVE PULMONARY DISEASE (COPD)	COPD is a broad category of lung problems including emphysema, chronic bronchitis, bronchiectasis, and chronic asthma in adults. A main component of all these diseases is the obstruction of inhalation and exhalation. COPD is responsible for more than 100,000 deaths each year and is the fourth leading cause of death in the United States.
CIRRHOSIS	Cirrhosis is extensive scarring and hardening of the liver. This condition is most often associated with advanced liver disease.

CORTICOSTEROIDS (STEROIDS, PREDNISONE)	A class of drugs modeled after hormones released by the body's adrenal glands. They are the most potent anti-inflammation drugs currently available and can be lifesaving to people with severe COPD and asthma, but they're also known for having serious side effects.
EMPHYSEMA	A lung disease that involves damage to the alveoli or air sacs in the lungs. In emphysema, the damaged air sacs do not deflate normally so breathing is harder. Lungs with emphysema may be slow to expel used-up air and unable to fill with enough fresh air to ensure an adequate oxygen supply to the body. In Alphas, the lungs actually become hyper inflated or enlarged, and the emphysema occurs mainly in the lower lungs since that is where most of the AAT-deficient blood flows. Smoking-related emphysema is usually in the upper lungs. An Alpha who smokes or has smoked may have emphysema throughout their lungs.
ESOPHAGEAL VARICES	Enlarged veins in the esophagus resulting from the increased pressure in the portal vein through which blood flows into the liver. This commonly occurs in cirrhosis.
FIBROSIS OF THE LIVER	The presence of scar tissue made of collagen within the framework of the liver tissue. When the liver is badly scarred, the organ will not function properly.
GENES	Genes are sections of DNA that determine specific human characteristics; 25,000 genes exist. Each parent gives you one gene that can alone, or in combination, result in certain characteristics. Genes also hold the instructions for making proteins, each of which has a different function in the body.
GENOTYPE	The human genome is a very long complex combination of gene sequences. The genotype is a description of the variation of the sequence of a particular gene. The specific change in an individual's alpha-1 gene sequence, known as a genotype, determines their specific characteristics, which is their phenotype.
HEPATITIS	Inflammation of the liver which can be caused by viruses, abnormalities of the immune system, and medications, as well as Alpha-1.
HEPATOMEGALY	Enlargement of the liver. In some cases, the liver can be felt below the rib cage.
HEPATOSPLENOMEGALY	Enlargement of the liver and the spleen.
HETEROZYGOTE/HOMOZYGOTE	Every cell of the body is composed of genes and every gene is actually a pair of alleles, one from the father and one from the mother. If your mother and father each give you the same allele, this gene is called a homozygote. If your mother and father each give you a different allele, this gene is called a heterozygote. Heterozygotes most often have one normal allele (M) and one abnormal allele (Z), a combination known as MZ. Alphas that are homozygotes have two abnormal genes, such as ZZ.
ICTERIC	Yellowing of the whites of the eyes associated with jaundice.
INFLUENZA	Commonly known as the flu, influenza is an acute, contagious viral infection, commonly occurring in epidemics. It is characterized by inflammation of the respiratory tract and by the sudden onset of fever, chills, muscular pain, headache and severe fatigue.
JAUNDICE	A condition characterized by a yellowish tint of the skin, white portion of the eye, tissue lining of the mouth, and body fluids due to excess bilirubin in the blood.
LIVER ENZYMES	Proteins (specifically enzymes) found in high concentration in the liver and lower amounts in the blood and body tissue. The enzymes are released into the blood when liver cells are injured. Doctors can measure the amount of enzyme released from cells and estimate the extent of liver damage using the AST (or SGOT), ALT (or SGPT), alkaline phosphatase, and GGT-P tests. There are other blood tests to monitor liver function that are commonly performed as well.

GLOSSARY OF TERMS continued

MICROMOLAR	Abbreviated as μM, it is used to designate the amounts of alpha-1 antitrypsin protein when serum levels are tested. A person is considered deficient in AAT protein when their serum level is 11 μM or below.
MILK THISTLE	This is an herb with an active ingredient called silymarin. Milk thistle is thought to be helpful for individuals with liver disease due to silymarin's cleansing and protective properties. It is extremely important that your healthcare provider be informed if you take, intend to take or intend to give this herb to your child. The National Institute of Health's National Advisory Council for Complementary and Alternative Medicine is currently studying this herb extensively.
PANNICULITIS	Panniculitis is an inflammation within the layers of fat beneath the skin which causes the skin to harden and form extremely painful lumps, patches, or lesions. It is likely that the damage is initiated by destructive action of unrestrained neutrophils. In some patients, damage from panniculitis may occur after an incident of trauma to the affected area. It occurs in children as well as adults, and has been linked to the ZZ and MZ phenotypes and possibly other alleles as well.
PHENOTYPE	The specific characteristic or type of ATT protein circulating in your blood; it is genetically determined by the alpha-1 genes received from your mother and father at birth. Other environmental factors may affect these characteristics.
PHLEGM	Thick, sticky, stringy mucus secreted by the mucous membrane of the respiratory tract, as during a cold or other respiratory infection.
PNEUMONIA	An acute or chronic disease marked by inflammation of the lungs and caused by viruses, bacteria, or other microorganisms and sometimes by physical and chemical irritants.
PORTAL HYPERTENSION	Blood flows from veins in the stomach, intestines, spleen and pancreas and goes into the liver through the portal vein. When the liver is diseased and unable to function properly, this blood flow is impaired, and pressure builds in the portal vein, which can cause a number of problems. This condition is known as portal hypertension.
PRURITUS	Medical term for itching.
SCLEROTHERAPY	A procedure that may be used in the treatment of bleeding from varices in the esophagus. Intravenous medication is injected directly into the enlarged veins to stop the bleeding.
SPLEEN	An organ that is a part of the lymphatic system in the human body. It functions as the body's defense mechanism, is involved in the formation and destruction of certain blood cells, and acts as a blood reservoir. Blood from the spleen goes into the liver.
SPLENOMEGALY	Splenomegaly, an enlarged spleen, occurs when the spleen has a disease or when portal hypertension develops due to liver disease.
SPUTUM	Matter coughed up and usually expelled from the mouth, especially mucus or pus that is expectorated (ejected or spit) in diseases of the air passages.
TPN	Total Parenteral Nutrition (TPN) is the administration of nutritionally-adequate solution intravenously; TPN may become necessary to provide nutrition to individuals with severe liver damage.
VITAMINS A, D, E, K	Fat-soluble vitamins that are necessary for proper nutrition and are frequently prescribed as dietary supplements when severe liver disease prevents their absorption into the blood stream.

WHERE DO I GET MORE INFORMATION?

LISTED BELOW ARE A NUMBER OF ORGANIZATIONS THAT HELP AND SUPPORT PEOPLE WITH ALPHA-1. EACH OF THESE ORGANIZATIONS WORKS WITH ALPHAS IN UNIQUE WAYS.

ALPHA-1 FOUNDATION

TOLL FREE: 1 (877) 2-CURE-A1 (228-732); WWW.ALPHAONE.ORG

The Alpha-1 Foundation is a not-for-profit organization dedicated to providing the leadership and resources that will result in increased research, improved health, worldwide detection, and a cure for Alpha-1 Antitrypsin Deficiency (Alpha-1). The Foundation provides the infrastructure to promote research and the development of new therapies for improving the quality of life of those diagnosed with Alpha-1. We are committed to close collaborations with medical experts, government agencies, international regulatory authorities, the pharmaceutical industry and other organizations to jointly resolve critical issues in the field of Alpha-1 research and treatment. A Grants Award Program supports a wide range of meritorious research in Alpha-1.

ABOUT THE ALPHA-1 ASSOCIATION

TOLL FREE: 1 (800) 521-3025; WWW.ALPHA1.ORG

The Association is a member-based not-for-profit organization helping to identify those affected by Alpha-1 Antitrypsin Deficiency, and to improve the quality of their lives through support, education and advocacy. The Association has a network of over 60 volunteer-led support groups throughout the United States.

ALPHANET

TOLL FREE: 1 (800) 577-ANET (577-2638); WWW.ALPHANET.ORG

AlphaNet assists patients and families with support, education, and strategies to manage their health. It also sponsors clinical trials for Alpha-1 therapies and produces *The Big Fat Reference Guide to Alpha-1* (BFRG), a complete guide to understanding, managing, and living with Alpha-1. (See inside cover page.) The BFRG also includes key terms, testing, genetics, and treatment options. It is available through the website or number listed above.

THE ALPHA-1 RESEARCH REGISTRY

TOLL FREE: 1 (877) 886-2383; WWW.ALPHAONEREGISTRY.ORG

The Research Registry is a confidential database of Alphas and carriers. The Registry gives patients the opportunity to provide information through questionnaires and clinical trials to help advance research on the disorder. It also provides access to experts on Alpha-1 care. Individuals enrolled in the Registry have the ongoing opportunity to participate directly in clinical trials of new therapeutic approaches in addition to other research opportunities.

AMERICAN ASSOCIATION FOR THE STUDY OF LIVER DISEASES

PHONE: 1 (703) 299-9766; WWW.AASLD.ORG

The leading organization of scientists and healthcare professionals committed to preventing and curing liver disease.

AMERICAN LIVER FOUNDATION

TOLL FREE: 1 (800) GO-LIVER (465-4837); WWW.LIVERFOUNDATION.ORG

The Foundation provides information on prevention, treatment, and potential cures of liver diseases.

AMERICAN LUNG ASSOCIATION (ALA)

TOLL FREE: 1 (800) LUNG-USA (586-4872); WWW.LUNGUSA.ORG

The ALA focuses on the prevention of lung disease including educational programs, research and advocacy.

CHILDREN'S LIVER ASSOCIATION FOR SUPPORT SERVICES

TOLL FREE: 1 (877) 679-8256; WWW.CLASSKIDS.ORG

This group serves the emotional, educational, and financial needs of families and children with liver disease.

ALPHA-1 KIDS

PHONE: 1 (410) 243-4499; WWW.ALPHA1KIDS.ORG

Alpha-1 Kids provides support and information for parents and children with Alpha-1.

CHOLESTATIC LIVER DISEASE CONSORTIUM

PHONE: 1 (303) 837-2598; WWW.RAREDISEASESNETWORK.ORG/CLIC

The Consortium provides support and information for children and families with rare cholestatic liver diseases. Go to the website and click on the Alpha-1 Antitrypsin Deficiency link for information on Alpha-1.

About the cover illustration: This original artwork depicts the passing of knowledge from mother to child. The scene represents the importance of informing others about Alpha-1.

ALPHAONE.ORG
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FORGING PARTNERSHIPS FOR A CURE

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ABOUT THE ALPHA-1 ASSOCIATION

Toll Free: 1 (800) 521-3025
www.alpha1.org

The Association is a member-based not-for-profit organization helping to identify those affected by Alpha-1 Antitrypsin Deficiency, and to improve the quality of their lives through support, education and advocacy. The Association has a network of over 60 volunteer-led support groups throughout the United States.

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