Addressing the need for providing up-to-date information about screening, diagnosis, and treatment of this disorder
**Foreword**

**Alpha-1 Antitrypsin Deficiency (AAT Deficiency)** is one of the most common serious hereditary disorders. AAT Deficiency has been identified in virtually all populations but is most common in individuals of Northern European (Scandinavian and British) and Iberian (Spanish and Portuguese) descent. Among patients with Chronic Obstructive Pulmonary Disease (COPD), up to 3% are predicted to have AAT Deficiency. It can also cause life-threatening liver damage in adults and children and liver cancer in adults. Despite its prevalence, patients and healthcare providers have been poorly informed about the disorder. For this and other reasons, the overwhelming majority of individuals with AAT Deficiency have not been detected. Of the more than 100,000 individuals in the United States estimated to have AAT Deficiency, less than 10% have been diagnosed, leaving more than 90,000 affected individuals undetected.

The discovery of AAT Deficiency by Laurell and Erickson in 1963 provided a foundation for current thinking about the pathogenesis of pulmonary emphysema. Although AAT Deficiency has become one of the best-understood genetic disorders at a molecular and cellular level, many questions about the clinical disorder remain unanswered.

The Alpha-1 Foundation, the National Institutes of Health (NIH) and pulmonary and liver disease experts are working aggressively to develop patient management and clinical treatment guidelines for working with patients affected by AAT Deficiency. This Healthcare Provider’s Guide to Alpha-1 Antitrypsin Deficiency is a response by the Alpha-1 Foundation to the need for providing up-to-date information about screening, diagnosis, and treatment of this disorder.

Materials in this document are designed to educate physicians, their staff members, and patients about Alpha-1 Antitrypsin Deficiency (AAT Deficiency) and the resources that are available for affected individuals, their family members and healthcare providers. An Educational Materials Working Group assists the Foundation’s Medical and Scientific Advisory Committee (MASAC) in identifying, producing, and reviewing educational and training materials. The Working Group is comprised of a wide range of professionals, including bioethicists, physicians, nurses and educators, all contributing their expertise. Individuals with AAT Deficiency are also included in the Working Group membership to add their personal expertise. Acknowledgment is made to all of these individuals and the many others who have provided insightful and helpful editorial comments. The Foundation also has educational materials for your patients, several available in multiple languages, including:

- What is Alpha-1 Antitrypsin Deficiency?
- A Guide for the Recently Diagnosed Individual
- What Does It Mean to Be an Alpha-1 Carrier?
- The Liver and Alpha-1
- Overview and Disorder Description
- What Is Alpha-1 Antitrypsin?
Deficiency. The S mutation is not associated with injury to the liver. As mentioned above, the S allele produces the S variant AAT protein and is associated with mild AAT Deficiency. However, the percentage of individuals with the S allele is only 10-15% of normal. When livers of these individuals are examined, hepatocytes often contain no AAT protein in the blood. Note that in Pi Z (ZZ), although these issues are still under investigation. Untargeted screening studies of large populations have revealed a variable prevalence of AAT Deficiency, depending upon the race and ethnicity of the study population. The family of the normal AAT alleles is referred to as M (or Pi M). The M alleles are the most common types of AAT gene and result in normal amounts and normal functionality of AAT in the blood. About 95% of the population of the United States has only M alleles. The rest are variants of the M allele (usually labeled M1, M2, M3, etc.).

Common Alleles:

- Pi S allele (S); (products from both genes don't appear to affect AAT levels or function. This aggregated AAT cannot be released from liver cells. As a result, the levels in the blood and tissues; and (2) deficient variants (those that don't appear to affect AAT levels or function). Generally, AAT genotypes are separated into two categories for diagnostic purposes. First, the result of many studies conducted worldwide. The presence of an unappreciated null allele. AAT in the serum can be found in the blood. "Pi" reflects the fact that AAT is a protein that is a component of polymorphonuclear leukocytes (neutrophils) and is shown in stainable form by isoelectric focusing and requires an experienced geneticist to interpret. AAT in the serum can usually be measured by a commercial test kit. AAT in the serum can be measured by a commercial test kit.

- Alpha-1 Antitrypsin Deficiency
  - What does it mean?
  - Antitrypsin Deficiency is a hereditary condition that affects the production of a protein called alpha-1 antitrypsin (AAT). AAT is produced in the liver and is involved in the body's defense against infection and inflammation. In individuals with AAT Deficiency, there is a deficiency of AAT, which leads to a reduced ability of the body to protect against lung and liver diseases. Alpha-1 Antitrypsin Deficiency is the leading genetic cause of liver disease in infants and children. In adults, it can lead to chronic lung disease and chronic liver disease.

- Diagnosis
  - Diagnosis of AAT Deficiency is usually based on a combination of clinical symptoms, family history, and laboratory tests. Laboratory tests may include a test for AAT levels in the blood and genetic testing to identify specific mutations in the AAT gene. Genetic testing can help identify the type and severity of AAT Deficiency and can guide treatment decisions.

- Treatment
  - Treatment for AAT Deficiency can vary depending on the severity of the condition and the underlying cause. Treatment options may include medications to reduce the symptoms of lung and liver disease, such as anti-inflammatory drugs, bronchodilators, and oxygen therapy. In some cases, lifestyle changes, such as quitting smoking and avoiding exposure to environmental triggers, may be recommended. In severe cases, liver transplantation may be considered as a treatment option.
Genetic Inheritance:

Genetic inheritance of AAT Deficiency follows simple Mendelian principles. Individuals with AAT Deficiency — could develop disease — could develop disease.

Moderate to severe deficiency

ZZ or SZ

Carrier

AAT Deficiency — may develop disease symptoms and does not carry any altered AAT genes. Pi MZ heterozygotes have one normal allele and one Z variant. They usually have normal or slightly decreased levels of AAT. They do not appear to be at an increased risk for disease, especially tobacco smoke. Counseling about avoidance of risk factors for lung and liver disease is recommended for all patients with AAT Deficiency.

Pi SZ individuals should also avoid potential risk for developing lung or liver disease, and does carry two altered AAT genes. Pi SZ heterozygotes have one allele for the S variant and one Z variant. They usually have normal or slightly decreased levels of AAT. They do not appear to be at an increased risk for disease, especially tobacco smoke. Counseling about avoidance of risk factors for lung and liver disease is recommended for all patients with AAT Deficiency.

Pi MZ individuals have one normal allele and one Z variant. They usually have decreased levels of AAT in their circulation; however, their levels can fall within the normal range. Although this issue remains under investigation, recent studies suggest that Pi MZ heterozygotes have a slightly increased risk for developing lung or liver disease compared to the general population with AAT Deficiency.

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Avoid both indoor and outdoor air pollution, especially in specialized urban regions and exposure to aerosolized particulates smaller than 10 µm (found in higher industrialized regions). It is also important to realize that pollutants and irritants in the environment can cause further irritation of the lungs and worsen the current condition of patients with AAT Deficiency. Smoking attracts neutrophils and macrophages to the lungs in large numbers and speeds the development of emphysema.

### 2. Avoid Environmental Pollution

#### 2.1 Smoking Cessation

Smoking is a major risk factor for lung disease in patients with AAT deficiency. Current smokers should stop smoking as soon as possible after diagnosis. Smokers are more likely to develop lung disease, even with AAT deficiency. Lifelong non-smokers will have a good chance of avoiding lung disease, even with AAT deficiency. Current smokers should stop smoking as soon as possible after diagnosis. Smoking is a major risk factor for lung disease in patients with AAT deficiency. Current smokers should stop smoking as soon as possible after diagnosis. Smokers are more likely to develop lung disease, even with AAT deficiency. Lifelong non-smokers will have a good chance of avoiding lung disease, even with AAT deficiency.

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#### 2.3 Avoid Infections

- **Respiratory infections**
  - Lung volumes
  - Spirometry (before and after inhaled bronchodilator)
- **Liver function tests**
  - ALT, AST, INR
  - Total and Direct bilirubin
  - Oximetry or arterial blood gases

#### 2.4 Avoid Occupational Exposures

Avoid occupational exposures to dust, fumes, and second-hand tobacco smoke from personal and second-hand tobacco smoke. These substances can cause further irritation of the lungs and worsen the current condition of patients with AAT deficiency. Smoking attracts neutrophils and macrophages to the lungs in large numbers and speeds the development of emphysema.

### 3. Development of an Exercise Program

#### 3.1 Supervised Aerobic and Strength Exercises

Routine exercise can improve mental outlook, stamina, and physical well-being. Exercise is essential to all aspects of patient care. The Alpha-1 Foundation is an important resource for patients with AAT deficiency. Case controlled studies have shown it is advisable for Alphas (persons with AAT deficiency) to avoid inhaled respiratory irritants and smoking tobacco. These substances can cause further irritation of the lungs and worsen the current condition of patients with AAT deficiency. Smoking attracts neutrophils and macrophages to the lungs in large numbers and speeds the development of emphysema.

### 4. Behavioral & Lifestyle Modification

#### 4.1 Avoid Infections

- **Respiratory infections**
  - Lung volumes
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- **Liver function tests**
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### Conclusion

In general, avoiding both indoor and outdoor air pollution, especially in specialized urban regions and exposure to aerosolized particulates smaller than 10 µm (found in higher industrialized regions) is important for patients with AAT deficiency. Smoking attracts neutrophils and macrophages to the lungs in large numbers and speeds the development of emphysema.
Medical Treatment

1. Vaccinations (influenza/pneumonia/hepatitis)
   - Yearly influenza vaccine and a pneumococcal vaccine at age 50 or older for those at risk.

2. Bronchodilators
   - Bronchodilators may be useful in relieving the symptoms of AAT Deficiency.
   - They can be administered as short-acting or long-acting medications.

3. Oxygen therapy
   - Oxygen therapy is recommended for individuals with severe airflow limitation.

4. Antibiotics
   - Prompt and aggressive treatment of infections is crucial for patients with AAT Deficiency.

5. Development of a Nutrition Program
   - Nutritional needs in those patients exhibiting liver damage should probably avoid alcohol completely.
   - Patients with any indications of AAT Deficiency-related liver problems, it may help to work closely with a nutritionist.
   - It is very important to inform the individual to carefully tailor or maintain good eating habits.
   - Since scientific research indicates that people with lung/liver disease need to consume more calories than "lung-healthy" people, this affects the manner in which your patient should approach nutrition.

6. Reducing Stress
   - The effects of stress and emotional responses are often overlooked or given little importance.
   - Here is a list of common symptoms they should be:
     - Fever
     - Chills with fever
     - Increase in quantity of phlegm or sputum
     - Darkening of color of phlegm or sputum
     - Coughing

7. Consultation on Stress Reduction Techniques
   - Relaxation techniques that help in reducing stress are often infectious or exposed to infections.
   - Avoid people who are sick (infected individuals).
   - Avoid children less than five years of age (who are more likely to get infections). The use of hand sanitizers is encouraged.
   - There are many techniques that help in reducing stress. Here are some:
     - Meditation
     - Breathing exercises
     - Yoga
     - Positive thinking
     - Hypnotherapy
     - Visualization
     - Biofeedback
     - Relaxation techniques
     - Belly breathing
     - Progressive muscle relaxation
     - Learning about the control of one’s stress response
     - Meditative activities
     - Relaxation training
     - Biofeedback training
     - Guided imagery
     - Improved exercise
     - Relaxation techniques and deep breathing

8. Developing a Pulmonary Rehabilitation Program
   - A Pulmonary Rehabilitation Program is highly recommended due to the increased neutrophilic infiltration seen in patients with AAT Deficiency.
   - This program includes education, smoking cessation, and nutrition retraining.

9. Exercise
   - Exercise is beneficial for improving lung function and endurance.
   - Improving sleep patterns can have a positive impact on mood.
   - Taking a walk (aerobic) is an excellent form of exercise, which may be beneficial in improving lung function and endurance.
   - Patients can benefit from aerobic activities such as walking, running, cycling, or swimming.
   - Some of the forms of exercise are:
     - Walking programs (particularly in climate controlled areas such as indoor shopping malls),
     - Strolling, swimming, and biking are all excellent forms of exercise, which may be beneficial in improving lung function and endurance.

10. Alcoholic beverages can damage the liver even in normal people. Many authorities recommend low, or no alcohol consumption for ZZ patients.

11. While an individual’s liver may not be affected, his/her nutrition may affect the liver.

12. Eating properly can help an individual with AAT Deficiency to maintain a healthy body weight, whether he/she has lung/liver disease or not.

13. Patients with AAT Deficiency (Alphas) report benefits from relaxation techniques that help in reducing stress.
Clinical Criteria for Use

1. Psychosocial/Family Support
   - The purpose of this section is to give you several scenarios that may arise when dealing with an AAT Deficiency diagnosis beyond a purely medical discussion, it may be helpful to review approach to talking with your patient about the issues specific to each person. There are no guarantees regarding the success at treating or improving any disease.

2. Employment
   - AAT Deficiency is the most common genetic cause of chronic liver disease, and the primary cause of liver failure in young people. The long-term outcome varies widely, from a few months to a lifetime of disease. Early detection and treatment can help prevent severe complications and improve survival. Regular check-ups and monitoring are important for individuals with low blood oxygen levels.

3. Reimbursement/Insurance claims
   - There are relatively few side effects reported: known are mostly rare. Augmentation therapy is not a cure; it will not reverse lung damage already present nor treat or prevent AAT Deficiency-related liver disease. This is not a treatment option for AAT Deficiency-related liver disease. Augmentation therapy cannot be recommended for AAT Deficiency-related emphysema. This is not a treatment for individuals with normal lung function. It is important to be aware of the manufacturing process and after packaging, there are minimal to no side effects. At various times during the manufacturing process, there are no side effects reported.

4. Corticosteroids
   - Numerous different types of surgery are available for those with end-stage lung disease from AAT Deficiency. These include miniature one-way valves and metal coils. It remains to be seen whether some people with AAT Deficiency might benefit from these procedures. Surgical ligation, are ineffective and the patient is not a candidate for a successful liver transplant. Each scenario is merely a starting point. Contacting the resources at the end of this guide will provide you and your patient(s) with more in-depth support and strategies for addressing each scenario that may arise.

Other Issues

5. Supplemental Oxygen
   - Oxygen therapy needs may be needed during exercise, with sleep, and, as oxygen levels decrease, at rest. Oxygen therapy needs are frequently assessed by the individual and his/her healthcare provider. For people who need supplemental oxygen, it has been shown to be life-prolonging. Oxygen can be administered via a nasal cannula, oxygen tent, or nasal prongs. Supplemental oxygen therapy is expensive and may be covered by insurance. It is recommended to consult with a healthcare provider to determine the appropriate type of oxygen therapy for each individual.
Insurance. Thus someone with AAT Deficiency, even
(emphysema, COPD, bronchiectasis, liver disease, insurance and employment, it does not cover life
“Your choice to be tested is totally voluntary. You can
While GINA prevents genetic discrimination in health
Establishing and maintaining confidentiality in the
Promoting Screening
and recommend that they familiarize themselves with
If your patient is currently uninsured:
However, benefits may vary depending upon where the
State laws concerning mandatory coverage.
Lifetime maximum benefits, if any. The Affordable
and reimbursement issues. If augmentation
insurance company regarding the use of augmentation
documentation requested from the patients by the
Foundation is to assist in the preparation of supporting
Each individual diagnosed with AAT Deficiency should
work in the future.

A: Yes. It is advisable to encourage your patient to

Q: Should I encourage my patient to discuss AAT

Q: Who will know the patient’s AAT Deficiency?

Q: Can your patient continue to work?

A: The results of the test will be included in a patient’s

A: The present state of your patient’s health

A: There is a minimal risk of an infection from obtaining

A: There may be some mild physical discomfort and

A: You may direct your patient to seek professional advice

A: Once the test results are back, I will ask you to come

A: Patients should inform future healthcare providers,

A: Instruct your patients to educate themselves regarding:

A: Note: Finding out about an ATT Deficiency diagnosis

A: If both parents are carriers, each child has a chance of

A: If your patient's physical condition does not allow

4. Genetic and other insurance claims

3. Funding

2. Education

1. Awareness

A: In the event of your patient's death, my agency,

A: Genetic discrimination policies and practices differ

“AAT Deficiency was discovered in 1963, there is still

A: Commonly, there are two conditions:

A: It shouldn't. Both GINA (the Genetic Information

A: Through a simple blood test, you can identify affected

A: Following are scenarios that were developed to assist

Q: Are you aware of any other environmental hazards

Q: Have your patients reported an episode of

Q: Who is at risk for AAT Deficiency?

Q: What happens if one parent is a carrier?

A: If your patient is currently insured:

A: You cannot refuse treatment based on AAT Deficiency.

A: 1. Health Insurance/Life Insurance

A: 2. Non-discrimination Act) and the Affordable Care Act

A: In the present state of your patient’s health
A. Explain the Recently Diagnosed Individual

After reviewing the results of the blood test we performed to determine if you had the genetic disorder AAT Deficiency, I must inform you that the results were positive for the disorder AAT you do have is slightly different from the normal antitrypsin (AAT) in your blood, and indicates that you do not have the disorder.

Consequences, including the genetic risk to the family, are discussed in the brochure “Am I an Alpha-1 Carrier?” which is available at the front desk. I would be pleased to go over this brochure with you. It is important to avoid all tobacco smoke, whether it is directly smoked tobacco products or situations where it is inhaled second-hand. At this point, you can begin discussing possible next steps, including your upcoming appointment to meet with a counselor to begin discussing the implications of your test results.

The progression of AAT Deficiency is difficult to say if your child will definitely need a liver transplant. The majority of children diagnosed with AAT Deficiency have a low rate of disease progression. Do you have any questions about your diagnosis or the genetic risk to your family?

B. Review information in the manual A Guide for the Parents of a Pediatric Patient with Liver Disease

If both parents are carriers, each child has a chance of inheriting AAT Deficiency, a chance of being a carrier (carriers) such as yourself, or have AAT Deficiency (two abnormal genes). Despite the negative result, you should still advise your blood relatives of the test result because of the possible risk. Carriers have one normal gene and one gene for the disorder. This combination of genes does NOT typically cause health problems. Currently, the risk of lung or liver disease in adults with Pi MZ. The health effects of cigarette smoke on the lungs can be magnified in those with Pi MZ.

C. Schedule the next patient visit

Discuss need for vaccinations

Discuss referring patient to a counselor

Discuss developing an exercise program

Discuss developing a nutrition program

Discuss requirement of follow-up visits

Avoid being around exposed individuals who are ill with the flu or a cold, etc.

Discuss need for baseline lung evaluation or screening

Discuss smoke any form of tobacco, including cigars, pipe, and/snus

Discuss alcoholic beverage consumption

Discuss risk of occupational and environmental exposures

Discuss surgery options (if appropriate)

Discuss baseline testing (with subsequent testing if appropriate)

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D. Complete the Treatment Checklist

- Explore options for counseling and support
- Obtain information and application for the Alpha-1 Research Registry
- Provide information and application for the Transplant Evaluation and Assistance Program
- Make sure that you note your counseling discussion in the record
- Discuss need for vaccinations
- Discuss referring patient to a counselor
- Discuss developing an exercise program
- Discuss developing a nutrition program
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support and answers to topics such as affected by AAT Deficiency and provides programs include:

- **Patient Information Line**
  - Toll Free: (1-800-245-6809) is available free to anyone with AAT worldwide. Some of the Foundation’s activities include genetic counseling, finding a cure, and supporting people with AAT Deficiency. Each of these activities is funded through donations, memberships, and the sale of books and other materials. To learn more, visit www.alpha1.org

- **Genetic Counseling Program**
  - Offered free phone-based testing options.

- **AlphaNet**
  - The Alpha-1 Foundation Clinical Resource Centers (CRCs), including the Alpha-1 Foundation Clinical Research Program (CRP), are clinical practices with particular expertise in AAT deficiency. The network is also comprised of over 80 volunteer boards, committees, and working groups.

- **Alpha-1 Research Registry**
  - Administers the Alpha-1 Coded DNA Tissue Program that provides confidential DNA and tissue samples for research studies. It is located at the University of Florida.

- **Alpha-1 Research Grants and Scholarships**
  - Supports research in the study of AAT deficiency. For more information, contact the Alpha-1 Foundation because of their interest in alpha-1-detection-program/

- **Targeted Detection Program**
  - Provides confidential testing for AAT Deficiency. See www.alpha1.org/alpha-1-test.html

- **Research**
  - Administers the Research Registry also administers the Alpha-1 Coded DNA Tissue Program that provides confidential DNA and tissue samples for research studies. The Medical University of South Carolina administers the Research Registry.

- **Public Policy and Advocacy**
  - Focuses on achieving results through research, prevention, and someday, a cure for this disease. The COPD Foundation was established to bring scientists together to focus on special topics related to AAT deficiency and experience in treating individuals with AAT deficiency. The COPD Foundation promotes research and provides information on COPD. The Foundation’s activities include prevention, treatment, and cure of COPD. The Foundation’s activities include genetic privacy and discrimination, and experience in treating individuals with AAT deficiency. The COPD Foundation promotes research and provides information on COPD. The Foundation’s activities include prevention, treatment, and cure of COPD.

- **Community Resources**
  - Provides a wide range of support services for AAT Deficiency. For more information, call (800) 465-4837.

- **Skinny Little Reference Guides**
  - Provides support for people with AAT Deficiency. To find a Support Group near you, visit www.liverfoundation.org

- **Big Fat Reference Guide to Alpha-1**
  - Provides support for people with AAT Deficiency. To find a Support Group near you, visit www.liverfoundation.org

- **Big Fat Reference Guide to Alpha-1**
  - Provides support for people with AAT Deficiency. To find a Support Group near you, visit www.liverfoundation.org

- **www.liverfoundation.org**
  - Web Site: www.liverfoundation.org

- **www.alpha1.org**
  - Web Site: www.alpha1.org

- **www.copdfoundation.org**
  - Web Site: www.copdfoundation.org
The Alpha-1 Foundation is committed to finding a cure for Alpha-1 Antitrypsin Deficiency and to improving the lives of people affected by Alpha-1 worldwide.

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