Alpha-1 antitrypsin deficiency

1. 0 INTRODUCTION Imagine you are walking through the park; it’s a warm day, you are surrounded by beautiful flowers, grass, and trees. Now imagine that these normal, everyday activities such as, walking outside on a humid day, make it more difficult for you to breathe. Instead of taking a normal breath, you are only able to inhale at thirty percent capacity. That is like breathing through the end of a tiny crimped straw. Also, imagine having to carry various inhalers and oxygen as part of an everyday routine. This is how it feels to have Alpha-1 Antitrypsin Deficiency, also known as early onset emphysema. For someone with Alpha-1 Antitrypsin Deficiency, everyday tasks can seem nearly impossible. Simply walking upstairs or raking leaves become extremely difficult chores. Alpha-1 Antitrypsin Deficiency is a genetic disorder that affects the lining of the lungs, making the body more susceptible to inhaled irritants and infections. As part of the family of Chronic Obstructive Pulmonary Diseases (COPD), often, Alpha-1 Antitrypsin Deficiency can be misdiagnosed as asthma or chronic bronchitis. In addition to lowered immunity, those with Alpha-1 Antitrypsin Deficiency have decreased lung capacity and various liver problems. The disease can be detected through a series of blood tests and other physical imaging scans. While there is currently no cure for Alpha-1 Antitrypsin Deficiency, there are known ways to minimize the symptoms allowing those with the illness to live a more typical lifestyle. 2. 0 HISTORY Alpha-1 Antitrypsin Deficiency was discovered in Sweden in 1963. Two physicians named Sten Eriksson, a clinician, and Carl-Bertil Laurrell, who led a clinical lab, ran different tests on individuals that were all diagnosed with chronic lung disease. After several long months of research, the two clinicians realized that the same enzyme, known as Alpha-1 Antitrypsin, was missing out of several of the patients. A

chemical analysis of the patients' blood showed that they were lacking Alpha-1 Antitrypsin, which was clearly a very important substance. The name of the disease originated from the serum antiprotease deficiency, also known as AAT. 3. 0 GENETICS Alpha-1 Antitrypsin Deficiency is characterized by the lack of a protective protein. Alpha-1 Antitrypsin is a protein made in the liver, which is then released into the bloodstream. This protein helps protect the lungs from inflammation caused by infection, tobacco smoke, and other inhaled irritants. Also, it plays an important role in preventing the breakdown of enzymes in various organs. In order to maintain proper levels of Alpha-1 Antitrypsin, an individual must receive two M (normal) genes from each parent. While an estimated 20 million people carry the recessive defective A1AT gene, only people who have two copies of this autosomal gene will be affected. The disease mostly affects Caucasians, with an estimated 1 in 2500 individuals diagnosed . MM genes, shown in 95% of Caucasians, are considered a normal phenotype. On the other hand, 95% of deficiency alleles in the general population have either S or Z. S allele less destructive and is associated with the milder lung diseases, where the Z allele is with severe liver and lung disease. 4. 0 ANATOMICAL AND PHYSIOLOGICAL Alpha-1 Antitrypsin Deficiency affects mostly the lungs and liver. The major features of the lungs include the bronchi, the bronchioles and the alveoli. The bronchi provide a passage for the air to move in and out of the lungs. The bronchioles are the smaller airway passages of the lungs that branch off from the bronchi and attach to the alveoli. The alveoli are the microscopic blood vessel-lined sacks in which oxygen and carbon dioxide gas are exchanged. The liver serves a wide variety of body functions, including detoxifying blood and producing bile that helps in digestion. Alpha-1

Antitrypsin Deficiency affects the lungs by slowly destroying the lung tissue enabling the lung to function correctly. The immune cells in the lung, whose job it is to prevent and fight infection, are also affected by cigarette smoke. They restrain the cells from clearing away bacteria and other particles. The inflammation caused by the immune system constantly attacking bacteria or tar leads to the release of destructive enzymes from the immune cells. Alpha-1 Antitrypsin Deficiency may also cause Cirrhosis of the liver. Cirrhosis is a chronic liver condition caused by scar tissue and cell damage. Cirrhosis makes it hard for the liver to remove toxins from the blood. These toxins build up in the blood and may affect brain function. 5. 0 SYMPTOMS There are many different types of symptoms that go along with Alpha-1 Antitrypsin Deficiency. The main symptoms are: · Shortness of breath - you may feel like you can't get enough air or your chest may feel tight. · Wheezing - a whistling noise in the chest during breathing, when the airways are narrowed or compressed. · Chronic cough — a cough that lasts longer than three weeks · Reoccurring chest colds · Jaundice - yellowing of the skin and eyes caused by too much bile pigmentation in the blood · Vomiting blood · Decreased exercise tolerance · Non-responsive asthma 6. 0 DIAGNOSIS In order to diagnose Alpha-1 Antitrypsin Deficiency blood work must be done. There are three types of blood work that can be done: · Alpha-1 genotyping — examines a persons genes and genotype. · Alpha-1 Antitrypsin PI — determines the type of Alpha-1 Antitrypsin protein present in a persons' blood. · Alpha-1 level test — determines the amount of Alpha-1 Antitrypsin in a persons' blood. To confirm the severity of the disease other tests may be done such as, chest x-rays and Computerized Automatic Tomography (CAT) scans. The chest x-ray examination may reveal a barrel-shaped chest. Also,

listening to the chest with a stethoscope may reveal wheezing, crackles, or decreased breath sounds. 7. 0 TREATMENT As far as treatments for Alpha-1 Antitrypsin Deficiency, there is currently no cure, but there are ways to relieve symptoms and to slow down the advancement of the lung disease. The first defense of treatment given to individuals with Alpha-1 Antitrypsin Deficiency is bronchodilators. Bronchodilators are medications used to relax the muscles around the airways, helping the airways to open up, making it easier to breathe. Bronchodilators are administered by a portable inhaler or by a nebulizer. Also there is a Proteinase Inhibitor, i. e. ProlastinÃ’, available that is used as a replacement therapy for the deficient Alpha-1 Antitrypsin. ProlastinÃ’ is administered intravenously. Methods used intravenously are a catheter inserted into a vein in the arm, or through a subcutaneous pre-inserted port. Therapeutic administration of ProlastinÃ’ is typically 60mg/kg body weight weekly. In some cases, an individual might develop end stage lung disease making a lung transplant necessary. 8. 0 WAY OF LIFE No one can completely understand what it is like to have Alpha-1 Antitrypsin Deficiency unless they experience it first hand. All my life I have grown up watching my father, Daniel Gift, suffer from this disease. Before he started showing symptoms of A1AT Deficiency, he was an active, hard working, daily exercising adult. Unfortunately he was a smoker, which increased the effects of Alpha-1 Antitrypsin Deficiency. It wasn’t until he was about 35 years old when he started showing signs of early onset emphysema. Within ten years of his diagnosis he was unable to continue working and could no longer be involved in outdoor activities with his family. In the summer my mom, sister, brother and I would go to the pool almost everyday coming home to my dad who was stuck in the house all day. The chlorine in the water and the hot sun

was too much for him to handle. When inhaled the chlorine causes an asthmatic attack. The effects of the medication prevent him from being in the sun. My brother was never able to go outside and throw the football or play catch with my dad because of excursion. Also, my dad has to circumvent contact with other people to avoid contracting potential colds and flu. 9. 0 CONCLUSION Alpha-1 Antitripsin Deficiency is a disease that affects many people throughout the world. There is currently no cure, but people have been treated with a combination of bronchial-expanding medications and changes in the way in which they live. Diagnosed through a series of tests, doctors not only know more about the illness, but also, are becoming more able to detect the disease. The disease affects both the lungs and the liver, which leads the person with the illness to make a series of lifestyle adjustments. People with Alpha-1 Antitripsin Deficiency must avoid potential airborne irritants and infections due to their diminished immune system. They must also limit their physical activity because breathing is more difficult with the disease. Alpha-1 Antitripsin Deficiency is a disease that is not only potentially devastating; it is also widespread and under diagnosed.