Need to talk? Call us 0845 2412173 Monday to Friday 9am to 5pm

Climb™

Supporting those affected by Inherited Metabolic Disorders

Alpha-1 Antitrypsin Deficiency

Alpha-1 Antitrypsin (AAT) Deficiency is caused by a defect in a gene known as SERPINA1 which leads to a deficiency or fault of the alpha-1 antitrypsin (AAT) protein. AAT is needed to control enzymes that are used to protect the liver and lungs and clear waste products away from the cell. Specifically it protects against an enzyme called neutrophil elastase which is released from white blood cells to fight infection. Due to the deficient or faulty AAT the enzyme can attack normal tissues, especially the air sacs in the lungs and causes lung disease. AAT Deficiency also means the body cannot remove waste from the liver cells and abnormal AAT can build up causing damage to the liver. Signs and symptoms of lung disease often develop between ages 20 and 50. AAT Deficiency affects between 1 in 3000 to 1 in 4000 in the UK. Exposure to factors such as tobacco smoke and dust can exacerbate symptoms.

Synonyms

Alternative names for this condition are:

- AAT
- AATD
- Alpha-1 Protease Inhibitor Deficiency
- Alpha-1 Related Emphysema
- Genetic Emphysema
- Hereditary Pulmonary Emphysema
- Inherited Emphysema

Further information about this condition is available from Climb.

Disclaimer

Please read our disclaimer and information on data protection.

Updated 12/05/17

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