AlphaNet When and Why Augmentation Therapy is Prescribed

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Alpha-1 Antitrypsin Deficiency (AATD) and Augmentation Therapy

- ✓ Alpha-1 antitrypsin deficiency (AATD) is a genetic condition. It is not a disease.
- ✓ Individuals with AATD (often referred to as "Alphas") have an increased risk of developing certain diseases. The most frequently occurring are lung disease and liver disease.
- ✓ If your physician has prescribed augmentation therapy or is considering it, it is important for you to understand which disease is being treated by this therapy.
- ✓ In most instances, augmentation therapy is used to treat a lung disease called emphysema. Emphysema is a form of chronic obstructive pulmonary disease (COPD). Chronic bronchitis, bronchiectasis, and asthma may co-exist with emphysema.
- ✓ Augmentation therapy uses alpha-1 antitrypsin protein from healthy plasma donors to supplement the levels circulating in the blood and lungs of Alphas. Therapy is infused into a vein, usually weekly.

Augmentation Therapy to Treat Lung Disease

- ✓ In Alphas, the shortage of alpha-1 antitrypsin protein allows an enzyme called neutrophil elastase to destroy air sacs in the lung called alveoli. Destruction of alveoli leads to the development of emphysema.
- ✓ The primary goal of augmentation therapy is to increase the alpha-1 antitrypsin protein level in the lungs. Boosting this level slows or stops lung damage, especially during exacerbations.
- ✓ Until a newer therapy is approved, augmentation therapy is considered a lifelong treatment.
- ✓ Alphas who use augmentation therapy to treat lung disease can still develop liver disease.
- ✓ It is controversial whether augmentation therapy should be continued following a lung transplant.

Should Augmentation Therapy Be Used in Healthy Alphas?

- ✓ Augmentation therapy should not normally be used in Alphas without lung disease because:
  - ❑ Not everyone with AATD will develop lung disease. Even Alphas with a genotype that is severely deficient will not necessarily develop lung disease.
  - ❑ Augmentation therapy is a time-consuming and expensive treatment that has potential side effects. Given that some individuals with AATD will never develop lung disease, it is not worth the expense, time, and risk to prevent the possibility of lung disease developing.
- ✓ Avoiding exposures such as smoking and dust/fumes in the workplace prevents lung disease.
- ✓ Annual lung testing is recommended to monitor lung health.
- ✓ Healthy individuals have more lung capacity than they need. It's important to pay attention to symptoms of shortness of breath with exercise. These symptoms help identify early emphysema, which will require a CT scan to diagnose. Asymptomatic individuals do not usually require augmentation therapy.

Augmentation Therapy to Treat Panniculitis

- ✓ While primarily used for emphysema, augmentation therapy is sometimes prescribed to treat panniculitis, a skin disease associated with inflammation of the panniculus, which is the fatty tissue beneath the skin. It is not explicitly approved by the FDA for panniculitis but is prescribed for this condition due to its efficacy.
- ✓ Higher doses of augmentation therapy are often required for panniculitis than for emphysema.
- ✓ When prescribed for panniculitis, augmentation therapy may only be needed during an outbreak of this skin disease.
- ✓ Alphas can have both emphysema and panniculitis.

Why Isn't Augmentation Therapy Used to Treat Liver Disease?

- ✓ Liver disease from AATD develops due to liver damage caused by misfolded alpha-1 antitrypsin proteins that have gotten trapped in the liver.
- ✓ Augmentation therapy does not affect the process through which alpha-1 antitrypsin proteins get trapped in the liver. Thus, it cannot be used to prevent or treat liver disease.
- ✓ There is no evidence that augmentation therapy helps—or harms—the liver.