Genetics Uncoded:

FACTS ABOUT ALPHA-1 ANTITRYPsin DEFICIENCY

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

What Your Test Results Mean

Testing results indicate that you are a carrier of alpha-1 antitrypsin deficiency. Carriers typically show no symptoms of alpha-1 antitrypsin deficiency; however, carriers are at an increased risk of having a child with alpha-1 antitrypsin deficiency. Risk for the current or future pregnancies is dependent on your partner’s carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

Alpha-1 Antitrypsin Deficiency Explained

Alpha-1 antitrypsin deficiency is an inherited condition that causes lung and liver disease. Lung symptoms including shortness of breath, fatigue, unintentional weight loss, and recurrent lung infections usually start to appear in adulthood. Many people with alpha-1 antitrypsin deficiency eventually develop emphysema, or damage to the alveoli. Liver disease can begin much earlier in affected individuals: it can cause jaundice as early as infancy, and some develop cirrhosis (scarring) of the liver in adulthood. Due to the scarring, people with alpha-1 antitrypsin deficiency have an increased risk for liver cancer. The severity of the disease can vary, and environmental factors such as exposure to tobacco smoke, chemicals, and dust can play a determining role. Individuals with the SZ or MZ allele combination are not as severely affected as those with the ZZ combination, but are at an increased risk for developing lung disease, especially if they smoke. To manage the disorder, smoking cessation, preventive vaccinations, bronchodilators, supplemental oxygen, and physical rehabilitation may be recommended. Intravenous enzyme replacement therapy benefits some patients.

How the Genetics Work

Alpha-1 antitrypsin deficiency is an autosomal recessive disorder caused by mutations in the SERPINA1 gene. In general, individuals have two copies of the SERPINA1 gene, one inherited from each parent. They may have any combination of three versions of this gene: M, S, or Z. Individuals with alpha-1 antitrypsin deficiency have two copies of the Z version of SERPINA1, while the individuals with MZ or SZ combinations are affected to a lesser extent. Those with MS or SS combinations are carriers typically unaffected by symptoms of the disease, and those with two copies of the M version are not considered to be carriers of the disease. Risk for two carriers to have a child with the disorder is 25%.

Recommended Next Steps

Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

Questions?

Contact us at (855) 776-9436 to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.