Clarity Genetics

Genetics Uncoded:

Facts about

Alpha-1 Antitrypsin Deficiency

What Your Test Results Mean

Carriers show no symptoms of alpha-1 antitrypsin deficiency and are not at risk to develop symptoms of the disorder. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

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 variants in the *SERPINA1* gene. In general, individuals have two copies of the *SERPINA1* gene, one inherited from each parent. All individuals will have a combination of a specific version of this gene, most commonly having a combination of M, S, or Z alleles, but other more rare alleles are also reported. Individuals with alpha-1 antitrypsin deficiency have two copies of the Z version (ZZ) of *SERPINA1*, while the individuals with MZ or SZ combinations are affected to a lesser extent. Those with MS or SS combinations are carriers and are not expected to develop symptoms of the disorder. Risk for two carriers to have a child with the disorder is typically 25%.

Questions?

Contact us at **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.

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