

You have access to a revamped Alpha-1 Alleles website (www.alpha1alleles.com), offering an improved platform for you to access information on specific variants of the Alpha-1 antitrypsin deficiency (Alpha-1, AATD, SERPINA1) gene.

Alpha-1 antitrypsin deficiency is a genetic condition. The Alpha-1 gene has over 100 variants. Some of these variants stop your body from producing enough normal Alpha-1. These gene variations are known as “alleles”.

The normal variant of the Alpha-1 gene is labeled “M”. Abnormal alleles, also called pathogenic variants or mutations, occur due to changes in the DNA of the Alpha-1 gene that affect how Alpha-1 is made. Mutations result in an abnormal quantity and/or abnormal function of alpha-1 antitrypsin. Two of the most common variants that increase your risk for lung or liver disease are labeled “S” and “Z”. There are also more than 100 other rarer deficiency alleles (such as F and I and null alleles). Since everyone has two of these genes (one from each parent), there are multiple combinations.

The Alpha-1 Alleles website (www.alpha1alleles.com) provides detailed insights into each allele variant or variant combination, including their contribution to Alpha-1 antitrypsin deficiency, biological mechanisms, effects on alpha-1 antitrypsin levels, and associations with lung and liver diseases. It also offers resources such as video tutorials for understanding Alpha-1 and navigating the website efficiently.

You can search for Alpha-1 alleles of interest via a basic dropdown menu or an advanced search option. While over 100 Alpha-1 alleles are currently reviewed on this site; there are additional alleles that have not yet been added, and novel variants that are likely to be discovered in the future. Additional alleles will be reviewed and added to this site on an ongoing basis.

It's important to note that the Alpha-1 Alleles website does not serve as a diagnostic tool or provide medical advice. Please consult your healthcare provider for personalized guidance.

For more information about Alpha-1 alleles visit: www.alpha1alleles.com. Additional resources can also be found on the Big Fat Reference Guide site: www.bfrg.alphanet.org.