Alpha 1 Antitrypsin MZ Information & Research

News & Research Update

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Website status and research update

Dear Subscribers,

This week's topic is the impact of Alpha1 on connective tissue, specifically on blood vessels.

Ehlers-Danlos Syndrome.

Some of you have been diagnosed with Ehlers-Danlos syndrome, a group of disorders that affect connective tissues supporting the skin, bones, blood vessels, and many other organs and tissues. Defects in connective tissues cause symptoms that range from mildly loose joints to life-threatening complications.

ELS is known to be a genetic defect and does not need to be related to an Alpha1 Antitrypsin deficiency. However, Alpha1 Antitrypsin deficiency is also known to have an impact on connective tissue, which can cause similar effects as described above, because Alpha1 Antitrypsin is important in remodeling and repair of the connective tissue.

Below is an extract from a paper with the link added;

“AAT inhibits elastase, which is important in the process of remodeling and repair of the connective tissue. Elastase production increases with inflammation, and an individual with an MZ or MS AAT genotype will have increased degradation of the connective tissue matrix, leading to decreased joint stability.”

Here is the link to the paper: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2911626/

Conclusion, when you are diagnosed with ELS, and the gene tests are not conclusive, you may experience the effect of Alpha1 Antitrypsin on your connective tissue.

Coronary Artery

Although slightly loose joints are considered an inconvenience, there are also life-threatening conditions caused by Alpha1 connective tissue issues, like when they affect the Coronary Artery.

A recent paper from 2024 states, “In patients with AATD, the lack of Neutrophil Elastase inhibition and overactivity of proteases can degrade elastin and connective tissues, leading to a loss of elasticity in vessel walls, increased stiffness, and reduced distensibility. This can result in aortic wall weakening and aortic distention. There is an abundance of evidence reporting the protective role of AAT in preserving arterial wall integrity.”

“Data show a pathological association between AAT and the development of aortic distention, with age as a driving factor. AAT is genetically highly polymorphic, with multiple alleles influencing serum levels and its functionality. A pathological association is evident between different AAT alleles with respect to vascular disease and the coexistence of emphysema, as well as Aortic and Cerebral Aneurysms. It is, therefore, important to consider the expanding scope of AATD, and patients with this condition should be monitored for cardiovascular implications.”

See this recent paper out of 2024; https://pubmed.ncbi.nlm.nih.gov/38283099/

In short, simple terms, your blood vessels will lose their flexibility and become stiffer / brittle, making them more easy to “crack.” Just think about an old garden hose, which snaps open because it loses its flexibility. It may be game over when this happens in your main blood vessels. A nice Alpha1 MZ member of our group told us that she has lost many (old) family members this way.

Conclusion

When you are getting older and you are an Alpha 1, it may be good to talk to your doctor about this and monitor for aortic and cerebral aneurysms on a regular basis.

Below is a figurative image of a blood vessel with an erupting aortic aneurysm.