Alpha 1 Antitrypsin MZ Information & Research

News & Research Update

Mar 23, 2024

Website status and research update

Dear subscribers,

This week, again, good progress related to our study towards the cause of our clinical conditions. In a report out of the UK Biobank, which contains 450.000 participants, we found that MZs have a much higher prevalence of biliary track issues, including a higher prevalence of nonalcoholic chronic liver diseases and a higher prevalence of abnormal liver functions. These results confirm and support our hypothesis and earlier study results. We will add these studies to our website in the following weeks.

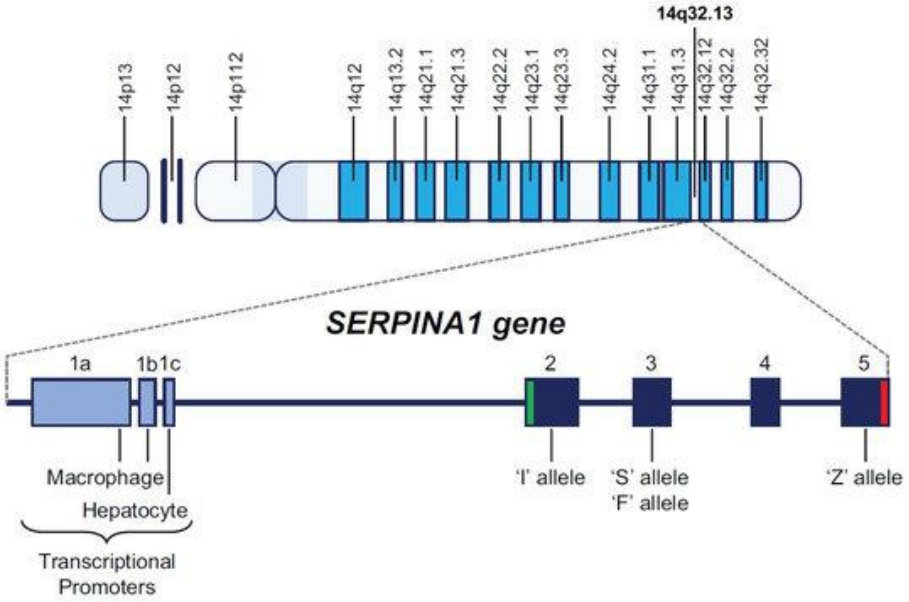
We also received a couple of questions from patients and medical professionals about interpreting the different mutation types of the SERPINA1 gene and how they relate to the AAT serum levels. To answer these questions, we have added two comprehensive reports from 2021 to the website. These reports provide an excellent, detailed overview of the mutations in the SERPINA1 gene, which encodes the alpha-1 antitrypsin (AAT) protein.

We also quickly summarized the most common mutation types, including combinations and AAT serum levels.

There are also many rare mutation types. If you are interested, you can find these in these two reports on the website.

Please note that you inherit a SERPINA1 gene from each of your parents. The SERPINA1 genes are codominant, which means that each SERPINA1 gene is responsible for producing 50% of the Alpha1 Antitrypsin.

Below is a picture of the SERPINA1 gene, showing the location of the different mutations and a picture of the various Alpha1 mutation variants.



Common SERPINA 1 Variants

SERPINA 1 Variant	Type of Mutation
Z	Point mutation (c.1096G>A;p.Glu366Lys)
S	Point mutation (c.863A>T;p.Glu288Val)
I	Point mutation (c.187C>T;p.Arg63Cys)
F	Point mutation (c.739C>T;p.Arg247Cys)

Alpha 1 Antitrypsin Serum Values for the most common variants

SERPINA 1 Variant Combination	Description	AAT Serum Level (mg/dl)
MM	This is the Normal "Healthy" Combination	126.9–159.2
MS	Intermediate deficiency genotypes	105.0–131.4
SS	Intermediate deficiency genotypes	88.2–107.5
MZ	Intermediate deficiency genotypes	75.8–96.6
ZF	Severe deficiency genotypes	64.5–79.2
SZ	Severe deficiency genotypes	46.4–61.0
ZZ	Severe deficiency genotypes	0–28.2