

# Alpha1 MZ Foundation - Information & Research

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## News & Research Update

Jul 20, 2024

Dear Subscribers,

We hear a lot that MZ's are not taken seriously by medical professionals. We can sympathize with that because most of us experienced the same when we were searching for answers and treatment for the medical issues we are experiencing.

In this issue, we would like to explain a bit more about what will be required for our group to be recognized and receive the appropriate treatment and resolve the disease burden among the 35 Million MZ's.

First of all, the terminology used in most research papers for Alpha1 with genotype MZ is "*carriers*". This is of course not correct because Alpha1 Antitrypsin deficiency is an inherited *co-dominant* condition, meaning that both alleles are responsible for 50% of the production of the Alpha1 Antitrypsin (AAT) protein.

This means that when you have a single mutated allele, (e.g. the "Z") you will produce less Alpha1 Antitrypsin, and the rest gets stuck in your liver which may cause liver damage, and you may also develop other symptoms/diseases because of your ~50% lower Alpha1 Antitrypsin status.

When you are a "ZZ" the diagnosis is quite clear, because your AAT level will be very very low and the majority of the AAT gets stuck in the liver. So no questions here, and there are sufficient medical papers providing proof that most ZZ's will develop lung emphysema over time, and that a part of the ZZ's will develop liver issues. (Depending of the autophagy speed controlled by a set of other genes) So the diagnosis by a medical professional in case of a ZZ is quite straightforward and will be accepted by insurance companies without questions asked.

For our large group of MZ's (35Mil worldwide), it's a different story.

Only in the last couple of years, more research became available related to the MZ group, where it has become evident that a single "Z" allele is sufficient to cause serious medical issues.

In most cases, an "MZ" who visits a medical professional, gets the story that when you don't smoke, and don't drink alcohol. You will be fine.

However, most of us know better, and we all know MZ persons in our group, who lived a healthy life, who did not smoke and still have lung emphysema and even lung transplants, and MZ's, who did not drink with serious liver issues.

We would also like to point out an obvious but important part of being an "MZ", and that is the fact that we still have the 'M' allele. This sounds good, (*more AAT*) but this also may have a negative effect on the liver, and as such, causes medical issues/symptoms which are hard to diagnose because they may not be directly related to the low AAT level, but towards a continues stress level of the MZ liver. Please note that in one of the research papers they mention that 25% of the MZ's have a higher background inflammation level. In other papers, we see clear links between MZ's and Cholestasis and Preeclampsia (just to name a couple) and there are a lot more issues that are typical for MZ's,

which indicates that the disease burden of an MZ may not be comparable to e.g. a ZZ and needs much more dedicated research and attention, especially because this is such a large group!!

It's up to us, as a group to take action...., and provide the medical proof (research papers) such that you and your MZ family (children and grandchildren) are recognized, properly diagnosed, and get the correct treatment.

This is why we are all part of the Alpha1 MZ Foundation, which has a clear focus on research specifically for the MZ population.

We as a group need to work together and we cannot stress enough that every individual contribution counts. Tell your story, educate yourself, and don't hesitate to ask questions.

We are preparing for a survey among our MZ members, and next week we have a call with a professional institution related to the implementation & execution of this study.

In summary, we are standing strong as a group, and the more MZ's are participating the stronger we as a MZ group will become to help you and your family!!

**And like always, enjoy the ride !!**

Our website <https://www.alpha1mz.org>

Facebook: <https://www.facebook.com/groups/790526129640939/>

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