

Alpha1 MZ Foundation - Information & Research

News & Research Update

Jul 6, 2024

Dear Subscribers,

In this issue:

1. Update on Beam Therapeutics
2. Update on the intermittent pain in the liver area we talked about last week
3. New very interesting item about the relationship between Autism and Alpha1 MZ

Beam Therapeutics

We reported about Beam Therapeutics in our June 15 newsletter. They now announced that the first patient was treated with BEAM-302, in their Phase 1-2 clinical trial.

This is a base editing therapeutics turning the Z into an M (You can look on our website for more information about this announcement) <https://www.alpha1mz.org/alpha-1-medication>

Intermittent pain in the liver area

Last week we talked about the “pain” in the right hypochondriac, which is very common in the Alpha1 MZ population, and the most likely cause is the biliary tract issue, which is common in MZ's. During discussions on this subject, a second but less probable cause of this “pain” was brought up, which is Perihepatitis. It is an uncommon, chronic manifestation of pelvic inflammatory disease affecting women of childbearing age. The classic manifestation of perihepatitis, or Fitz-Hugh–Curtis syndrome, is severe right upper abdominal pain that often radiates to the shoulder, and can be diagnosed directly via visualization of adhesions between the diaphragm and liver, or liver and the anterior abdominal wall.

Conclusion is that there are two possible causes, but considering the prevalence of the biliary tract issues within our group the most likely source is the biliary tract. When it is really getting painful, go to an MD to check it.

Autism and Alpha1 MZ

A paper out of 2009, about this subject was brought to our attention. (Thank you !)

“Low Serum Alpha-1 Antitrypsin (AAT) in Family Members of Individuals with Autism Correlates with PiMZ Genotype” <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2716675/>

In this paper they state that a significantly high number of autistic family members had lower than normal serum levels of AAT when compared to controls.

“Autistic children with regressive onset had significantly lower levels of AAT compared to controls, and a significant number of autistic children with low serum AAT also had hyperbilirubinemia, gastrointestinal disease and respiratory problems. We also found that a significantly high number of these individuals had the PiMZ genotype and correspondingly low levels of serum alpha-1 antitrypsin.”

This is a good proper study which clearly shows a relationship between autism and Alpha1MZ. Interesting that they also found bile acid issues (induced by the Alpha1 liver) and the corresponding issues in the gastrointestinal tract.

Another paper (not related to Alpha1) out of 2010 states that Hepatocyte Endoplasmic Reticulum stress is related to Autism, which basically confirms the findings of the paper out of 2009, because we all know that the major issue in the MZ population is caused by the hepatocyte ER stress.

What we don't know yet is the actual mechanism between Alpha1 MZ and Autism. In the last paper they speculate that autism may be the result of abnormal membrane trafficking of the synaptic functional molecules induced by ER stress. (Synapses are places where neurons connect and communicate with each other)

I personally feel that this has to be researched in much more detail, because the relationship with Alpha1 is not really known, recognized / mentioned in the medical world as far as I am aware of.

And like always, enjoy the ride !!

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