

Alpha1 MZ Foundation - Information & Research

News & Research Update

Sep 14, 2024

Dear Subscribers,

In this issue, we want to update you on the effects of Alpha1 Antitrypsin deficiency on the skin.

Did you know that the skin is the body's largest organ and comprises seven layers with a specific function? First of all, the skin is part of the body's immune system and acts as the body's first barrier against germs, ultraviolet light, chemicals, and injury. The skin also helps maintain body temperature and prevent water loss from the body. The first five layers of the skin are called the epidermis, the outer layer of skin that protects the body from infections, dehydration, and injury. It also renews cells in the skin. The next two layers comprise the dermis, the layer beneath the epidermis that contains blood vessels, nerve endings, hair follicles, sweat glands, and tiny muscles. The dermis provides elasticity, firmness, and strength to the skin.

We already know that an Alpha1 Antitrypsin deficiency affects inflammation, making it last much longer and, in general, much more intense. So, it will be harder to resolve when an inflammation affects your skin. For example, we heard quite some MZs about eczema, which makes perfect sense because Eczema (Atopic dermatitis) is a condition where inflammation is due to an imbalance of serine proteases and their naturally occurring inhibitors. So, having an Alpha1 Antitrypsin deficiency will make you more susceptible to Eczema. The paper referenced below is not recent but proves that the augmentation of Alpha1 Antitrypsin plasma resolved the eczema of patients with Eczema.

<https://pubmed.ncbi.nlm.nih.gov/1456482/>

Another known skin issue among Alpha1 Patients is Panniculitis, an inflammation of the subcutaneous fat (under the skin). When many inflammations are present and not treated correctly, this condition can even be lethal..! We actually know of an Alpha1 who died from Panniculitis because the NHS denied Alpha1 Antitrypsin augmentation treatment for this person in the UK.

Just so that you all know and remember, the only current suitable remedy/medication for these skin conditions is Alpha1 Antitrypsin augmentation therapy. So, your weekly doses of alpha-1 antitrypsin protein derived from the blood of healthy donors to increase your amount of AAT.

Below is an Abstract of a recent paper of 2022, which describes Alpha1-related Panniculitis

Alpha-1 antitrypsin deficiency associated panniculitis

Background: Panniculitis represents a rare and potentially lethal manifestation of alpha-1 antitrypsin deficiency (AATD). Evidence regarding management is limited to case reports and small case series. We sought to clarify typical features and investigation of AATD-associated panniculitis and assess the evidence regarding therapeutic options.

Findings: We identified 117 cases of AATD-associated panniculitis. AATD was present in 15% of all cases of biopsy-proven panniculitis. Failure to achieve clinical response was seen in all instances of systemic steroid use. Dapsone, although practical and accessible, is frequently associated with failure to achieve remission. In these instances, intravenous AAT augmentation therapy generally resulted in a response.

Conclusions: AATD may be more prevalent among patients presenting with panniculitis than previously thought. Patients presenting with panniculitis and systemic illness show a high mortality risk. Although most cases are associated with the severe ZZ-genotype, moderate genotypes may also predispose to panniculitis. Dapsone remains the most cost-effective therapeutic option, whereas intravenous AAT augmentation remains the most efficacious. Finally, glucocorticoids appear ineffective in this setting. You can find the link to the full paper below.

<https://www.jaad.org/action/showPdf?pii=S0190-9622%2821%2900232-2>

And, like always, enjoy the ride !!

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