



Case Study – Alpha-1 Anti-Trypsin Deficiency

Names and other identifying information have been changed to protect patient confidentiality.

Madison, 24 year old female

Madison was a heavy cigarette smoker. Her ActX test results showed that she had a SERPINA1 variant associated with a moderate form of alpha-1 anti-trypsin deficiency. Alpha-1 antitrypsin (AAT) helps protect the lungs from damage caused by smoking. Patients with her variant who are smokers often will get severe emphysema at a young age, in their 30's or 40's.

Outcome

Once Madison was presented with this finding, Madison, with her physician's assistance, was able to find the motivation to quit smoking, something she had previously failed at. By quitting smoking, she most likely has avoided developing severe emphysema.

(Note: Different variants in the SERPINA1 gene lead to moderate or severe AAT deficiency. In the case of the more common moderate AAT deficiency, which Madison has, non-smoking patients generally do not develop emphysema. In cases of severe AAT deficiency, emphysema will often develop in non-smokers, although smoking does aggravate the condition.)

Additional information on Alpha-1 anti-trypsin deficiency

AAT regulates neutrophil elastase, an enzyme that fights infection. Lack of or a reduction in function of AAT can result in neutrophil elastase attacking normal tissues, especially in the lung.

References

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