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Severe Alpha-1 Antitrypsin (AAT) deficiency is a hereditary condition caused by mutations in the SERPINA1 gene, which predisposes to lung emphysema and liver disease. It is usually related to PI*Z alleles, and less frequent to rare and null (QO) alleles. Null-AAT alleles represent the end of a continuum of variants associated with profound AAT deficiency and extremely increased risk of emphysema.

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