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# Modernising case finding for $\alpha_1$-antitrypsin deficiency by DNA sequencing of COPD patients

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Alpha-1-antitrypsin deficiency (AATD) is a hereditary metabolic disorder caused by mutations in the SERPINA1 gene that result in a reduction in the serum concentration of the protein alpha-1-antitrypsin (AAT), and a predisposition to COPD [1]. AAT functions as an inhibitor of neutrophil elastase (and other proteases) and is essential in maintaining a balance of protease and antiprotease activity in the lungs [2]. Imbalance causes an escalating cycle of inflammation and degradation of lung tissue that, over time, leads to COPD [3].