Alpha-1 antitrypsin deficiency (AATD)

Record Status
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Citation

Authors' objectives
Alpha-1-antitrypsin deficiency (AATD) is a genetic disorder that occurs due to abnormalities in the production of alpha-1-antitrypsin (AAT). The most common clinical conditions associated with AATD are chronic obstructive pulmonary disease (COPD) and liver disease. AAT is encoded by the serpin peptidase inhibitor, clade A (alpha-1 antiprotease, antitrypsin), member 1 (SERPINA1) gene with the normal variant M and 2 main deficiency variants Z and S. The Z variant is associated with severe AATD. The number of carriers of AAT deficiency variants is estimated to be 116 million worldwide, and severe AATD is estimated to be present in 1.1 million individuals. The highest prevalence of AATD is observed in individuals of European ancestry. AATD is diagnosed based on clinical criteria, including presence of COPD or liver disease, in addition to use of laboratory methods such as estimation of AAT levels, molecular genetic testing, and protein testing. In most laboratories, these testing methods are used in combination with one another either simultaneously or in series to improve accuracy of AATD diagnosis. The current report evaluates evidence for AATD testing using methods that detect Z and S variants, including polymerase chain reaction (PCR)-based methods and isoelectric focusing (IEF). Various applications of S and Z variant and phenotype testing are assessed, including presence of these variants and phenotypes in patients with COPD or liver disease, population screening, prenatal diagnosis, and augmentation therapy response.

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