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AAT. Alpha-1 Antitrypsin Deficiency

NIHR BioResource – Rare Diseases study project

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V1.1 16/05/2019

Summary

Alpha-1 Antitrypsin Deficiency (AATD) is an inherited condition associated with lung and liver damage in the form of emphysema and cirrhosis.

Not all patients with AATD develop disease, but we do not know why this is – genetic factors and influences like smoking may play a part. In addition it is unclear which medical investigations are essential to monitor disease or what aspects of disease are most important to people with AATD. Emphasis is often put on physical tests that patients report being difficult and uncomfortable to do yet these tests may not be the best for monitoring their disease.

By working with the NIHR BioResource we aim to identify blood and genetic markers that may relate to lung and/or liver disease severity and disease progression, with a view to developing a more efficient way of monitoring patients in the future, personalising this according to their risk of disease.

Recruitment Criteria

Inclusion

Any patient who has alpha 1 antitrypsin deficiency (AATD), as defined by 2 abnormal (non M) alleles on genotyping for AATD.