Micropathology Ltd

Please forward this flyer to your local biochemistry, respiratory, liver and genetics services who may be interested in this service

Molecular genetic diagnosis for alpha1-antitrypsin deficiency

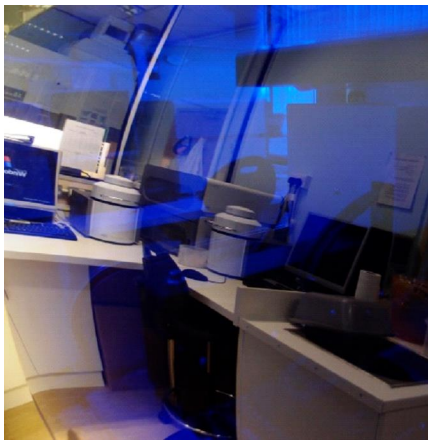
- We are pleased to announce the latest development in our real-time PCR assays for human genetics: **Analysis for common mutations that underlie alpha1-antitrypsin (A1AT) deficiency**

Clinical background

- Deficiency of A1AT may cause lung and/or liver disease. Lung symptoms in people with A1AT deficiency often develop before age 40 beginning with shortness of breath following light exertion, persistent symptoms of recurrent respiratory infection or asthma that do not respond to treatment. Without management symptoms progress to chronic obstructive pulmonary disease (COPD). About 10% of infants with A1AT deficiency present with neonatal jaundice which may progress to cirrhosis and liver failure.
- A1AT deficiency affects around one in 2000 people of Northern European ancestry.
- Early diagnosis of A1AT deficiency enables proactive clinical management and better patient outcomes. As the phenotype in the early stages of disease are non-specific a rapid, non-invasive molecular genetic test is a valuable diagnostic tool.

Target mutations

- The A1AT protein is encoded by the *SERPINA1* gene (previously referred to as *Pi* for protease inhibitor). Two specific sequence variants of *SERPINA1* are associated with A1AT deficiency, namely c.1096G>A (traditionally referred to as *PiZ*) and c.863A>T (traditionally referred to as *PiS*).
- People who have two copies of these gene variants, either ZZ, SS or SZ will have deficiency of A1AT in their serum and are at risk of developing symptoms.
- People with one copy of either S or Z and a normal allele have low levels of A1AT but rarely develop clinically significant disease.



Service Provided

- Our test is fully validated and EQA quality assured.
- We provide rapid reporting with a maximum 5 day reporting time.

For further information, please visit our website at www.micropathology.com

telephone 02476 323 222 or email Dr. Sarah Ball: s.ball@micropathology.com

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