ALPHA-1 Association of Australia

Information Sheet for your Physician
Testing for Alpha-1 Antitrypsin Deficiency

Alpha-1 Antitrypsin Deficiency (AATD) is under-recognised, taking an average of ~8 years between initial patient presentation and diagnosis. Yet it's estimated that around 1 in 9 Australians carry a defective Alpha-1 Antitrypsin (AAT) gene. AATD is often misdiagnosed as asthma or bronchitis.

AATD can cause disease of the liver (eg. cirrhosis, cancer), lungs (eg. early onset lower lung emphysema), sometimes skin (panniculitis), and is known to be associated with a range of other conditions. AATD can manifest as emphysema in adults as young as 30 years, sometimes younger. AATD is a significant cause of liver transplant in children.

When should a patient be tested?

- If a family member is known to have a defective AAT gene.
    - Early detection of high risk profiles enables patients to make informed risk-reducing lifestyle choices for themselves and their children, eg. avoid pollutants such as smoking or inner city living, avoid excess alcohol intake.
- If a patient presents with ongoing or repetitive respiratory symptoms.
- If a patient presents with unexplained liver complaints.
- If a patient presents with a skin rash that hasn't responded to treatment.

Tests required

MBS item 66635 - Quantitation in serum, urine or other bodily fluids.

> AAT levels can be affected by inflammation, trauma or malignancy, causing levels to temporarily increase by up to 25%. A marker for inflammation should be simultaneously tested, eg. C-reactive protein (**MBS item 66500**).

If AAT levels are 1.10 g/L or less:

MBS item 66638 - Isoelectric focussing or similar methods for determination of alpha-1 antitrypsin deficiency phenotype in serum.

Genotyping or DNA sequencing may be necessary to correctly identify AAT defective alleles other than the more common S and Z types.

For more information visit alpha1.org.au

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