7.08 Evaluation of AAT phenotyping in patients with low alpha-1 antitrypsin levels

Maqbool Moeez[1], Tomás P. Carroll[2], Ronan Heeney[2], Elaine Hayes[1].

1.Our Lady of Lourdes Hospital, Drogheda

2. Alpha-1 Foundation Ireland, Beaumont Hospital

Alpha-1 antitrypsin deficiency (AATD) is a hereditary condition that leads to low levels of an important antiprotease. The main function of Alpha-1 antitrypsin (AAT) is to protect the lungs, with deficiency leading to risk for lung, liver and skin disease. According to the HSE National Laboratory Handbook (1), phenotyping is recommended if AAT levels are low (<1.0 g/L) (1). It is also recommended that C-reactive protein (CRP) is performed in conjunction, as AAT is an acute phase reactant.

We performed a retrospective review of samples sent for AAT testing from Our Lady of Lourdes Hospital to determine compliance with testing guidelines.

We reviewed 83 AAT test results with AAT levels <1.0g/L, sent from our laboratory. Excluding duplicates, only 18 (25%) out of 72 individuals were phenotyped. 13/18 (72%) had clinically significant AAT deficiency. CRP was performed in only 37% of samples.

Patients who have been identified with low levels of AAT and who have not had phenotyping will be recalled for testing. A new protocol for AAT testing has been proposed with Beaumont Hospital and the RCSI Alpha-1 Foundation Ireland laboratories to ensure phenotyping of patients with AAT levels less than 1 g/L. This will lead to a more rapid diagnosis of suspected AATD cases and earlier intervention.

References

1. National Laboratory Handbook HSE, Ireland. www.hse.ie/eng/about/who/cspd/ncps/pathology/resources/lab-testing-for-alpha-1-antitrypsn-antibodies.pdf

Conflict of Interest: *None to declare*