JOURNAL CLUB

MUC5B and interstitial lung disease

In this study, the data for 2633 participants from the Framingham Heart Study were analysed to explore any association between a polymorphism in the promoter of a mucin gene (MUC5B) and radiological evidence of interstitial lung abnormalities.

Data for participants were evaluated to include physical examination findings, respiratory questionnaires, spirometry including diffusion capacity for carbon monoxide, genotyping for MUC5B and thoracic CT evaluation. Participants with the MUC5B variant were significantly more likely to have radiological interstitial lung abnormalities. Seven percent of CT scans examined demonstrated evidence of interstitial lung abnormalities—these participants were observed to have reduced total lung capacity, reduced diffusion capacity and were twice as likely to describe shortness of breath and a chronic cough.

This study was largely robust in its methodology and conclusions. The patient population (Framingham Heart Study) were however predominantly of European descent. Rates of radiological interstitial lung abnormalities were much higher in this cohort than previously observed rates of idiopathic lung fibrosis. It is thought that not all of these lung abnormalities will progress to idiopathic lung fibrosis. There is also likely a degree of underdiagnosis of idiopathic lung fibrosis.

Idiopathic lung fibrosis is a condition of significant morbidity and mortality, and although there are known associations with advancing age and smoking, the exact mechanism of disease is largely unknown. Twenty percent of the population have been found to have the MUC5B variant, although a far smaller proportion of the population display features of idiopathic lung fibrosis. This points to the fact that the disease is multifactorial in aetiology and further research is needed into the genetics of the condition and potential environmental triggers.[1]

▸ Hunninghake G, Hatabu H, Okajima Y, *et al*. MUC5B Promotor polymorphism and interstitial lung abnormalities. *N Engl J Med* 2013;368:2192–200.

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