



Mucin 5B promoter polymorphism is associated with idiopathic pulmonary fibrosis but not with development of lung fibrosis in systemic sclerosis or sarcoidosis

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Résumé en anglais

BACKGROUND: A polymorphism (rs35705950) 3 kb upstream of MUC5B, the gene encoding Mucin 5 subtype B, has been shown to be associated with familial and sporadic idiopathic pulmonary fibrosis (IPF). We set out to verify whether this variant is also a risk factor for fibrotic lung disease in other settings and to confirm the published findings in a UK Caucasian IPF population. **METHODS:** Caucasian UK healthy controls (n=416) and patients with IPF (n=110), sarcoidosis (n=180) and systemic sclerosis (SSc) (n=440) were genotyped to test for association. The SSc and sarcoidosis cohorts were subdivided according to the presence or absence of fibrotic lung disease. To assess correlation with disease progression, time to decline in forced vital capacity and/or lung carbon monoxide transfer factor was used in the IPF and SSc groups, while a persistent decline at 4 years since baseline was evaluated in patients with sarcoidosis. **RESULTS:** A significant association of the MUC5B promoter single nucleotide polymorphism with IPF ($p=2.04 \times 10^{-17}$; OR 4.90, 95% CI 3.42 to 7.03) was confirmed in this UK population. The MUC5B variant was not a risk factor for lung fibrosis in patients with SSc or sarcoidosis and did not predict more rapidly progressive lung disease in any of the groups. Rather, a trend for a longer time to decline in forced vital capacity was observed in patients with IPF. **CONCLUSIONS:** We confirm the MUC5B variant association with IPF. We did not observe an association with lung fibrosis in the context of SSc or sarcoidosis, potentially highlighting fundamental differences in genetic susceptibility, although the limited subgroup numbers do not allow a definitive exclusion of an association.

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