Genetic variant link with long-term incidence of interstitial lung disease in rheumatoid arthritis

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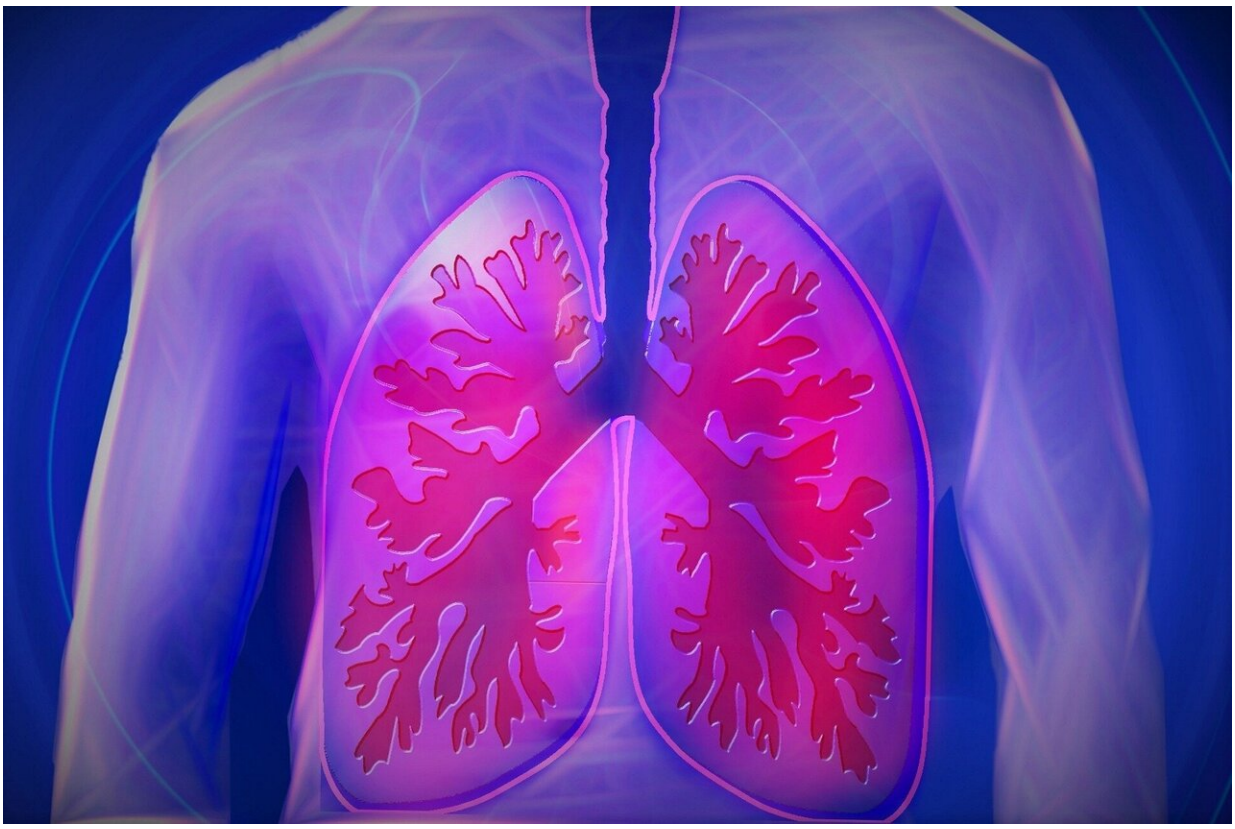

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Rheumatoid arthritis (RA) is an inflammatory autoimmune disease that causes pain, swelling and stiffness in the joints. It can also cause fatigue,

and the underlying inflammation may affect other body systems. Up to 10% of people with RA are affected by interstitial lung disease (ILD) during their lifetime, and ILD is one of the leading causes of death in patients with RA. Data presented at the 2021 EULAR congress report findings from a longitudinal study showing that people with MUC5B gene variant have a considerable lifetime risk of ILD, and that this gene variant contributes to increased morbidity. These findings have clinical implications for improving identification of people with RA who are at high risk for developing ILD.

The MUC5B gene codes for mucin—a protein that is normally found in mucus secretions, and which is part of the body's natural defence against infection. The promoter variant called rs3570595 0is a common variant in the MUC5B gene, with an allele frequency of 0.1 in the Finnish population. Overexpression of MUC5B in lungs influences the development of pulmonary fibrosis. The promoter variant rs35705950 in MUC5B is the strongest known genetic risk factor for rheumatoid arthritis-associated interstitial lung disease (RA-ILD). However, there are no large-scale data on the impact of the MUC5B promoter variant on the long-term incidence of RA-ILD.

Antti Palomäkiand colleagues used FinnGen—a collection of epidemiological cohorts and hospital biobank samples—to describe the long-term risk of RA-ILD in people with RA carrying the MUC5B promoter variant compared to those without the variant. FinnGen is able to link people's genetic information with up to 46 years of follow-up data within nationwide registries.

Of 248,400 people, 5534 had been diagnosed with RA, and 178 of these (3.2%) had developed ILD. The MUC5B promoter was a strong predictor of developing ILD in people with RA, conferring a lifetime risk of ILD of 14.5% by age 80, compared to 5.2% in people with RA who did not carry the promoter variant. In the general population of

people without RA, MUC5B promoter carriers and non-carriers had lifetime risks of developing ILD of 3.9% and 1.3%, respectively.

The authors found that the risk difference started to emerge at the age of 65. The risk was highest in men with RA who are MUC5B carriers. In this group,18.5% of carriers developed ILD, compared to 8.5% of non-carriers.

These findings have clinical implications for improving identification of people with RA who are at high risk for developing ILD.

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