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## GENETIC VARIANT LINK WITH LONG-TERM INCIDENCE OF INTERSTITIAL LUNG DISEASE IN RHEUMATOID ARTHRITIS

**A population biobank study of 250,000 individuals in Finland**

**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 rs35705950 is 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 Palomäki and 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.

### Source

Palomäki A, et al. *MUC5B* promoter variant and long-term incidence of interstitial lung disease in patients with rheumatoid arthritis: a population biobank study of 250,000 individuals. Presented at EULAR 2021; abstract OP0007.

### About EULAR

EULAR is the European umbrella organisation representing scientific societies, health professional associations and organisations for people with rheumatic and musculoskeletal

diseases (RMDs). EULAR aims to reduce the burden of RMDs on individuals and society and to improve the treatment, prevention and rehabilitation of RMDs. To this end, EULAR fosters excellence in education and research in the field of rheumatology. It promotes the translation of research advances into daily care and fights for the recognition of the needs of people with RMDs by the EU institutions through advocacy action.

### **About the EULAR European Congress of Rheumatology**

Since its introduction in 2000, the annual EULAR European Congress of Rheumatology has become the primary platform for exchange of scientific and clinical information in Europe. It is also a renowned forum for interaction between medical doctors, scientists, people with arthritis/rheumatism, health professionals and representatives of the pharmaceutical industry worldwide. The EULAR congress is usually held in June in one of the major cities in Europe ([see previous congresses](#)).

The [scientific programme](#) at the congress covers a wide range of topics on clinical innovations, clinical, translational and basic science. Meetings set up by associations of people with arthritis/rheumatism, health professionals and the health care industry complement the programme. The poster sessions, offering lively interaction between presenters and participants, are regarded by many as the heart of the congress.

Over the years, the EULAR Congress has gained a reputation of being a most innovative platform for the practicing physician particularly with respect to the acquisition of information on novel clinical research. The congress attracts more than 18,000 delegates from more than 130 countries.

The aim of the EULAR European Congress of Rheumatology is to provide a forum of the highest standard for scientific, both clinical and basic, educational, and social exchange between professionals involved in rheumatology, liaising with patient organisations, in order to achieve progress in the clinical care of people with rheumatic diseases.

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