Uncovering genetic links to the development of pulmonary disease

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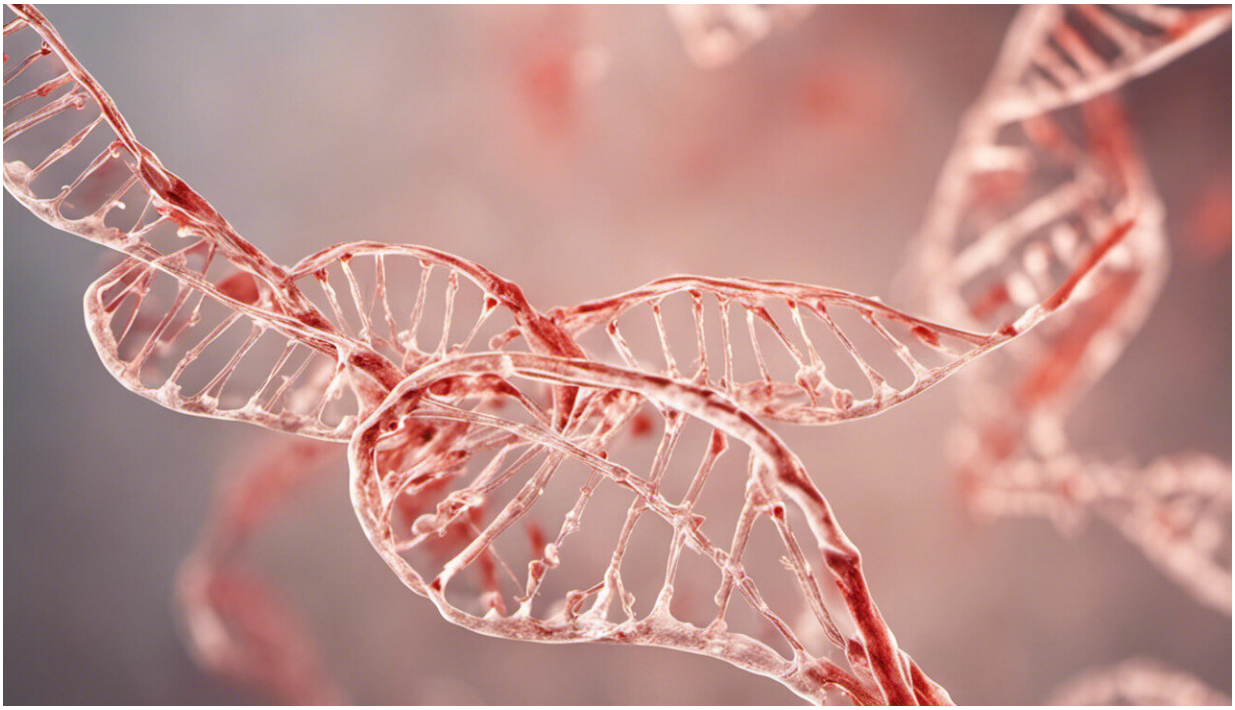

Credit: AI-generated image (disclaimer)

Building on EU-funded research, scientists have identified genetic traits that heighten the risk of developing chronic obstructive pulmonary disease.

Chronic obstructive pulmonary disease (COPD) is an incurable

progressive lung condition that kills over 5 million people every year. While smoking remains the single most important risk factor, genetics also clearly plays a key role; only one in four smokers are likely to develop COPD.

Understanding why some people are more predisposed to developing COPD than others is important because it could lead to more effective diagnoses and treatments. For example if identified early, genetic risk factors can be used as biomarkers, and high risk individuals advised to avoid smoking to prevent the onset of COPD.

Genetic breakthrough

Scientists recently made a significant breakthrough in this direction. Building on some of the pioneering findings of the EU-funded COPACETIC project, an international team of researchers carried out a comprehensive genomic analysis and were able to identify 13 new genetic regions associated with COPD. In addition, they also discovered four genetic regions that were not previously associated with any lung function trait.

An overlap between genetic risk of COPD and two other lung diseases – asthma and pulmonary fibrosis – was found. These discoveries will enable scientists to identify high risk individuals and focus on new biological pathways to deliver therapies for patients with this disease.

'These findings would only be possible with the kind of large collaborative efforts that supports this study. Not only do the results build on our knowledge of COPD, but also reveal potential links with other lung diseases, like pulmonary fibrosis and asthma and can form the underpinnings of a precision medicine strategy for the treatment of more than one lung disease,' said Dr. James Kiley, Director of the Division of Lung Diseases of the National Heart, Lung, and Blood Institute (NHLBI)

of the National Institutes of Health (NIH) in the US.

Building on knowledge

With scientific progress a continual process of building upon previous discoveries , these advancesin the field of COPD and genetics grew from important ground work carried out by COPACETIC. In this project, a consortium of researchers from the Netherlands, Denmark, Germany, Sweden and Poland conducted a genome-wide scan of individuals at high risk, collecting genetic material from thousands of smokers and non-smokers from across Europe.

Genome-wide association scans (GWASs) for COPD found approximately 350 DNA variations that were subsequently examined. Studies were also carried out to identify genes involved in chronic mucous hypersecretion and factors including, but not limited to, genetics leading to lung function decline. Baseline studies showed that COPD resulted from airflow obstruction or tissue damage, but not both.

These international efforts to better understand the genetics behind COPD have shown that while smoking remains the number one causal factor (and that stopping smoking is vital if COPD patients hope to get better), cessation on its own may not be enough to stave off the disease. While it is clear that genetics does play a role in who develops the disease, the task now is to find efficient ways of using biomarkers to identify those individuals, and to develop targeted therapies.

More information: Project website: www.copacetic-study.eu/english

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