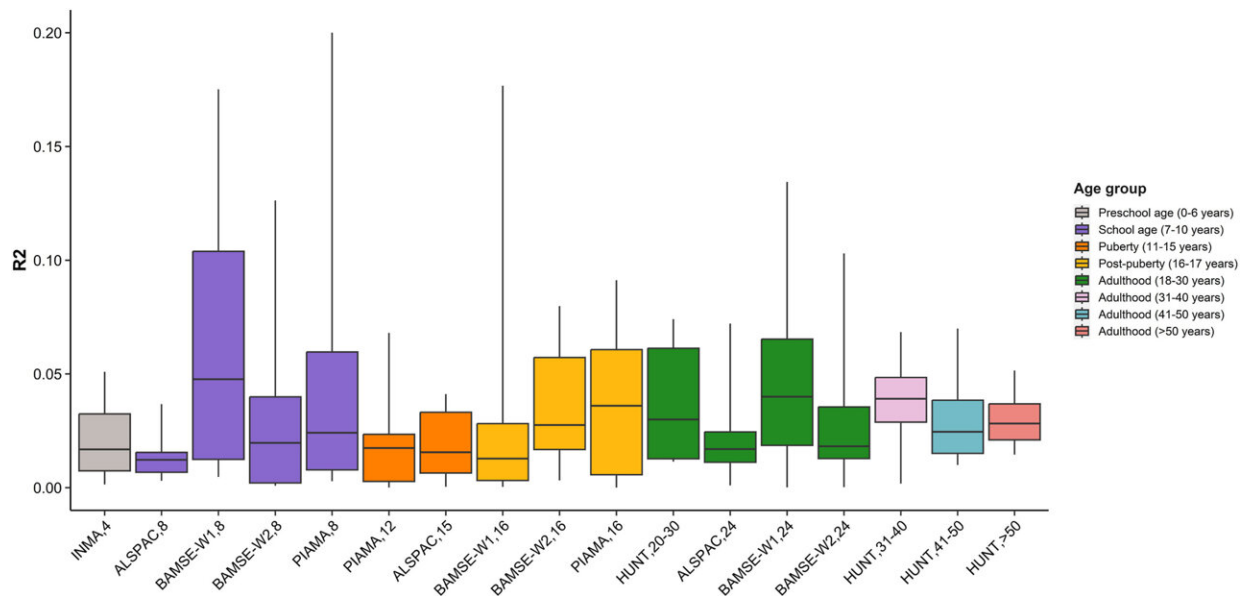


Genetics of COPD important for lung function in young people, study finds

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Box plot of variance in FEV1/FVC explained by the PRS for airflow limitation across the lifespan. Credit: *eClinicalMedicine* (2024). DOI: 10.1016/j.eclinm.2024.102731

Certain genetic variants that have previously been linked to the lung disease COPD can explain reduced lung function already in children and adolescents, according to researchers at Karolinska Institutet. The [results](#) are presented in the journal *eClinicalMedicine*.

More than half a million adults in Sweden and 300 million globally live

with [chronic obstructive pulmonary disease](#), COPD. The disease is characterized by impaired [lung function](#) and symptoms such as shortness of breath and mucus cough.

An important risk factor is smoking, but exposure to air pollution, recurrent respiratory infections, and heredity also have an impact.

In the largest study to date in the field, the researchers show that genes linked to COPD are also largely responsible for reduced lung function—and that this can already be seen in children.

"The fact that we have been able to show in a robust and objective way that COPD genes also affect lung function already in childhood is a paradigm shift. I hope it will mean that we can not only identify people at risk in time but also help to wash away the old 'stigma of COPD', as we now know that the disease process can begin early in life," says Erik Melén, professor of pediatrics at the Department of Clinical Science and Education, Södersjukhuset, Karolinska Institutet, who has led the study.

Preliminary results from the Swedish BAMSE study indicated that genetics were more important than previously thought for lung function, such as the genes that affect how the airways and the lung's supporting tissue are formed.

In order to be able to study a larger group of individuals, collaboration was initiated with 15 European centers that included nearly 112,000 participants who were followed over time. In these people, the researchers determined 82 genetic variants that had previously been linked to COPD in adults.

In more than 45,000 participants between the ages of 4 and 50, lung function was also measured.

"It was then possible to see that the link between genetic variants of COPD and impaired lung function was already strong in children from preschool age. These results were not affected by smoking, gender, or whether you had an asthma diagnosis," says Natalia Hernandez-Pacheco, researcher at the same department and the study's first author.

Hoping to prevent COPD

The researchers calculated a so-called polygenic risk score, PRS, for COPD, which could specifically explain just over six percent of the variation in lung function. They emphasize that it is the combined effect of many genetic variants that can be linked to reduced lung function.

One limitation is that the study participants are from Europe. The results can therefore not be applied to other groups.

The next step is to study the possibility of preventing the onset of COPD in people with an increased genetic vulnerability.

"For example, we want to investigate whether it is possible to carry out measurements of lung function in children, for example in school health care, in the same way that height and weight are measured today. It is also important to target preventive efforts in lung health towards those who are at increased risk of developing COPD in the future," says Melén.

More information: Natalia Hernandez-Pacheco et al, Exploring the genetics of airflow limitation in lung function across the lifespan – a polygenic risk score study, *eClinicalMedicine* (2024). [DOI: 10.1016/j.eclinm.2024.102731](https://doi.org/10.1016/j.eclinm.2024.102731)

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