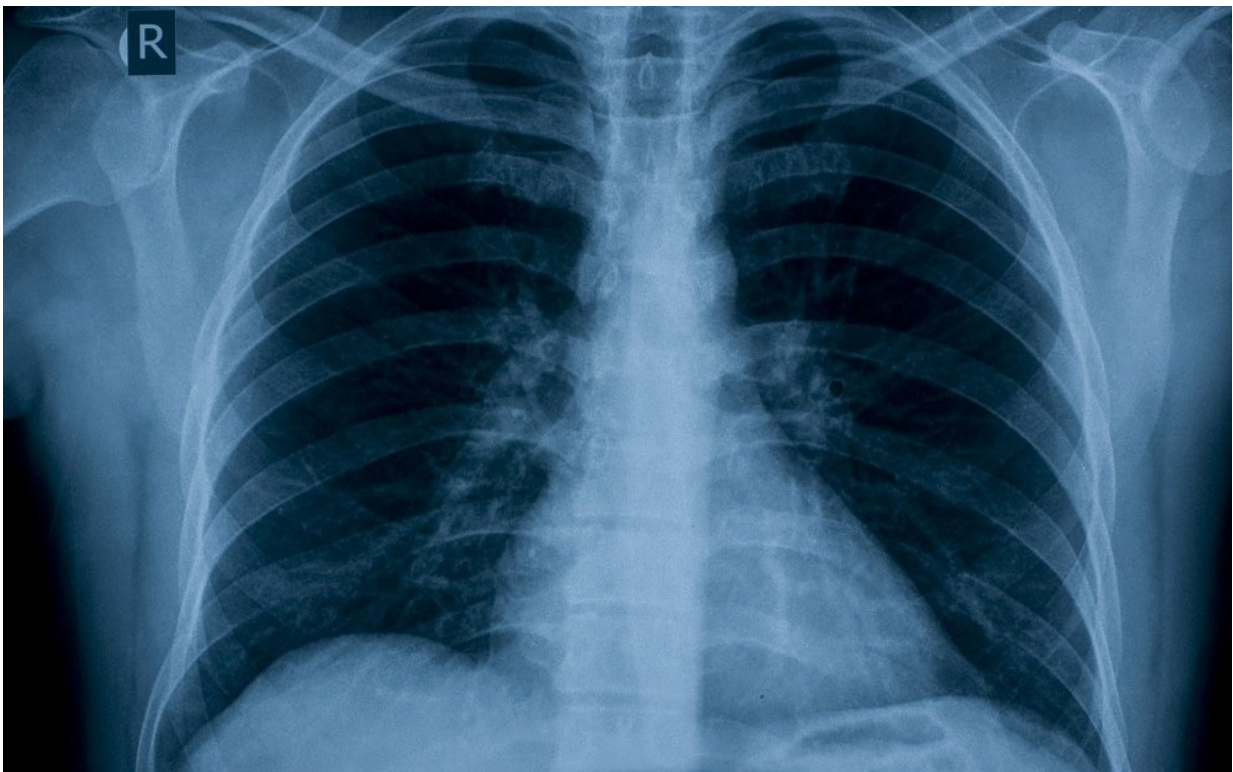


# Genetic study could lead to new treatments for sufferers of pulmonary arterial hypertension

April 13 2018

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Credit: St. George's University of London

Research carried out by a consortium including St George's, University of London has identified new genes for pulmonary arterial hypertension, which provides renewed hope for people affected with this incurable

condition.

Pulmonary arterial hypertension is a rare form of [high blood pressure](#) that specifically affects the blood vessels of the lungs. The [pulmonary arteries](#) become thickened and narrow, preventing normal blood flow and leading to increased [blood](#) pressure. Patients experience shortness of breath, fatigue, chest pain and fainting, as the heart struggles to deliver oxygen to the body. It has a poor prognosis without treatment, usually resulting in death from heart failure two to three after diagnosis.

A UK and Europe-wide consortium of researchers carried out whole-genome sequencing of 1,038 patients in the largest genetic study to date into this condition. They identified three novel [genes](#) and provided independent validation of another gene as a key risk factor in the disorder. Their study has been published in *Nature Communications*.

The identification of new genes relating to the condition highlights previously unexplored cell pathways in the disease's progression and opens up new avenues for potential drug discovery and development.

Dr Laura Southgate, Lecturer in Genetics at St George's University of London and a senior co-author of the research, said: "We knew that PAH has a strong genetic component but isolated cases were much harder to characterise genetically. This research takes us several steps closer to improved diagnosis and treatment as well as providing a comprehensive dataset for future researchers."

**More information:** Stefan Gräf et al. Identification of rare sequence variation underlying heritable pulmonary arterial hypertension, *Nature Communications* (2018). [DOI: 10.1038/s41467-018-03672-4](https://doi.org/10.1038/s41467-018-03672-4)

Provided by St. George's University of London

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