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Scientists discover new gene associated with debilitating lung disease

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Health scientists at the University of Leicester and University of Nottingham have heralded the discovery of a gene associated with lung fibrosis as 'a potential new avenue of treatment for further research into this terrible disease.'

The breakthrough is announced in a paper published in *The Lancet Respiratory Medicine*.

Idiopathic Pulmonary Fibrosis (IPF) is a debilitating lung disease, affecting ~6,000 new people each year, where scarring (fibrosis) of the lungs makes it difficult to breathe.

IPF, on average, results in death 3 years after diagnosis. There is no cure for IPF, and currently available drugs can only slow the disease down, and do not stop, or reverse, it. Furthermore, some patients may suffer unpleasant side-effects. A better understanding of the disease is needed to develop even more effective treatments.

Researchers Professor Louise Wain from the University of Leicester and Professor Gisli Jenkins from the University of Nottingham were lead authors of the study. They analysed the DNA from over 2700 people with IPF and 8500 people without IPF from around the world and found that people with IPF are more likely to have changes in a gene called AKAP13.

The researchers were also able to show that these DNA changes affect

how much AKAP13 protein is produced by the gene in the lungs. Researchers know from other studies, that AKAP13 is part of a biological pathway that promotes fibrosis (or scarring) and importantly that this biological pathway can be targeted with drugs. Taken together, the findings suggest targeting this pathway with drugs in people with IPF might lead to new treatments. To confirm this, the research team now need to undertake more detailed studies into the role of AKAP13 in people with IPF.

The work was led by researchers at Leicester and Nottingham and brought together collaborators from around the world to form the largest combined analysis of people with IPF undertaken to date.

Professor Wain, GSK/British Lung Foundation Chair in Respiratory Research at the University of Leicester, said: "We urgently need new ways to treat this terrible disease. Our findings highlight a potential new avenue for treatment and we now need more research to identify why this gene is important in IPF and how we can use that information to identify new therapies."

Professor Gisli Jenkins, University of Nottingham, said: "What is really exciting about these studies is that this gene affects a pathway that can be targeted by drugs currently in development, opening the door to precision medicine in IPF."

Ian Jarrold, Head of Research at the British Lung Foundation, said: "IPF is a condition with no known cause or cure and we urgently need to change this. Which is why funding for further research and campaigning for greater awareness is so crucial. This study is exciting and demonstrates that there is hope. We look forward to seeing how it develops."

Steve Jones, Chair of Action for Pulmonary Fibrosis, said: "This is

important research, which will give hope to the 33,000 people in the UK living with IPF and their families. We need more research like this into the genetic factors underlying the disease and possible treatments."

More information: *The Lancet Respiratory Medicine* (2017). DOI: 10.1016/S2213-2600(17)30387-9

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