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New genes for lung disease discovered

New genetics study by consortium led by universities of Leicester and Nottingham

Scientists have discovered five genetic variants that are associated with the health of the human lung. The research by an international consortium of 96 scientists from 63 centres in Europe and Australia sheds new light on the molecular basis of lung diseases.

The new findings provide hope for better treatment for lung diseases like Chronic Obstructive Pulmonary Disease (COPD) and asthma. In the past it has been difficult to develop new treatments because the molecular pathways that affect the health of the lung are not completely understood. It's hoped the new pathways discovered could in the future be targeted by drugs.

The ground-breaking research involved a genetic study of 2.5 million sites across the human genome involving samples from 20,000 people across the world. The consortium was led by Dr Martin Tobin from the University of Leicester and Professor Ian Hall from The University of Nottingham.

The research, part-funded by the Medical Research Council (MRC) and Asthma UK, is published today in Nature Genetics. It represents a significant advance because it is the first time that these five common genetic variations have been definitely linked with lung function.

The scientists said: "This work is important because until now we have known very little about the genetic factors that determine an individual's lung function. By identifying the genes important in determining lung function, we can start to unravel the underlying mechanisms which control both lung development and lung damage. This will lead to a better understanding of diseases such as chronic obstructive pulmonary disease (COPD) and asthma. Crucially, it could open up new opportunities to manage and treat patients with lung conditions".

The authors added: "A large reduction in lung function occurs in chronic obstructive pulmonary disease (COPD), which affects around 1 in 10 adults above the age of 40 and is thought to be the fourth most common cause of death worldwide. Smoking is the major risk factor for development of COPD. Lung function and COPD cluster within families, indicating that variations in genes also predispose individuals to reduced lung function.

"The scientists of the SpiroMeta consortium compared genetic variants at each of 2.5 million sites across the human genome in over 20,000 individuals of European ancestry with their lung function measures. In five different locations in the human genome, genetic variants resulted in alterations in lung function. The scientists showed that these were real findings by checking the effects of the same variants in over 33,000 additional individuals. They also compared their results to those of a second consortium, CHARGE, which has published a paper in the same issue of the journal.

The scientists emphasise that they do not expect these findings to lead to immediately to genetic tests to predict who will develop lung disease. What is more important, they say, is that the findings will help understand the underlying causes of lung diseases and thus may indicate new ways of treating the condition.

"The research would not have been possible without the generous support of the participants of the contributing studies from the UK, Europe and Australia, to whom we offer our thanks."

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NOTE TO NEWSDESKS

Lung function is commonly expressed using two measures recorded using a simple device called a spirometer. These measures are termed the FEV1 (or forced expiratory volume in 1 second) which is the volume of air that can be breathed out in 1 second, and the FVC (forced vital capacity) which is the total volume of air that can be breathed out. In chronic obstructive pulmonary disease (COPD), which encompasses chronic bronchitis and emphysema, narrowing of the airways causes a disproportionate reduction in FEV1. Cough, phlegm and shortness of breath are common symptoms of COPD. The simplest way to diagnose COPD is through spirometry, which is usually available in general practitioners' surgeries. Although there is no cure for COPD, stopping smoking and treatments can improve symptoms and reduce the impact of COPD on exercise and daily activities. Drug treatments include bronchodilators and, for exacerbations, may include short-term steroids. Patients with COPD are more susceptible to serious lung infections, so flu vaccination each winter is important.

The genetic determinants of COPD can be studied by investigating the genetic variants that affect the risk of developing COPD itself or by studying lung function itself, on which the diagnosis of COPD is based. Reduced lung function may also occur in patients with other airway diseases such as asthma.

Further research will be needed to study in detail the molecular alterations in the lung that result from the genetic variants identified, and to investigate whether these might be targeted by drugs. At this time there is no case for testing for common genetic variants that might predispose to COPD.

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Further information on lung function and on COPD and asthma is available from:

The British Lung Foundation:

The British Lung Foundation works for everyone affected by lung disease. The charity focuses its resources on providing support for people affected by lung disease today; and works in a variety of ways (including funding world-class research) to bring about positive change, to improve treatment, care and support for people affected by lung disease in the future.

It provides information via its website www.lunguk.org and telephone helpline on 08458 50 50 20

Asthma UK

Asthma UK is the charity dedicated to improving the health and well-being of the 5.4million people in the UK with asthma. For more information visit asthma.org.uk.

About the Medical Research Council

For almost 100 years the Medical Research Council has improved the health of people in the UK and around the world by supporting the highest quality science. The MRC invests in world-class scientists. It has produced 29 Nobel Prize winners and sustains a flourishing environment for

internationally recognised research. The MRC focuses on making an impact and provides the financial muscle and scientific expertise behind medical breakthroughs, including the first antibiotic penicillin, the structure of DNA and the lethal link between smoking and cancer. Today MRC funded scientists tackle research into the major health challenges of the 21st century. www.mrc.ac.uk

The University of Nottingham is ranked in the UK's Top 10 and the World's Top 100 universities by the Shanghai Jiao Tong (SJTU) and Times Higher (THE) World University Rankings.

More than 90 per cent of research at The University of Nottingham is of international quality, according to RAE 2008, with almost 60 per cent of all research defined as 'world-leading' or 'internationally excellent'. Research Fortnight analysis of RAE 2008 ranks the University 7th in the UK by research power. In 27 subject areas, the University features in the UK Top Ten, with 14 of those in the Top Five.

The University provides innovative and top quality teaching, undertakes world-changing research, and attracts talented staff and students from 150 nations. Described by The Times as Britain's "only truly global university", it has invested continuously in award-winning campuses in the United Kingdom, China and Malaysia. Twice since 2003 its research and teaching academics have won Nobel Prizes. The University has won the Queen's Award for Enterprise in both 2006 (International Trade) and 2007 (Innovation — School of Pharmacy), and was named 'Entrepreneurial University of the Year' at the Times Higher Education Awards 2008.

Nottingham was designated as a Science City in 2005 in recognition of its rich scientific heritage, industrial base and role as a leading research centre. Nottingham has since embarked on a wide range of business, property, knowledge transfer and educational initiatives (www.science-city.co.uk) in order to build on its growing reputation as an international centre of scientific excellence. The University of Nottingham is a partner in Nottingham: the Science City.

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- Ranked top with Cambridge for student satisfaction amongst full time students taught at mainstream universities in England
- Ranked as a Top 20 UK university by the Sunday Times, Guardian,Times and UK Complete University Guide, published in The Independent
- Ranked in world's top 3% of universities by Shanghai Jiao Tong International Index, 2005-08 and the Times Higher Education-QS World University Rankings
- Students' Union of the Year award 2005, short listed 2006 and 2007

This is Leicester: http://www2.le.ac.uk/about/facts