

Manchester researchers world first in complex genetics testing

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A grandfather-of-three from Tameside is helping University of Manchester researchers become the first in the world to assess all the genetic links with Chronic Pulmonary Aspergillosis (CPA) - a debilitating fungal lung infection that claims the lives of thousands of people in the UK and millions more worldwide.

Using state of the art next generation exome* sequencing technology, 61-year old Doug Steele will join 160 other patients from across the UK to have his DNA assessed and compared for all the individual gene links.

Despite more than 20 years of illness which has left him with only 40 per cent lung function and in need of almost daily drug injections, Doug, who lives in Dukinfield, says he was very happy to sign up for the study. "I hope it helps me of course I do, but I know it will help others. I have utmost faith in the team who through their treatment and care have saved my life."

New [diagnostic tests](#) and opportunities for novel treatments to prevent progressive lung destruction by the airborne fungus *Aspergillus* are likely discoveries. The work will take about 18 months to complete and will cost £150,000.

Leading the research team is Dr David Denning, Professor of Infectious Diseases in Global Health, at The University of Manchester, who explains: "CPA causes progressive destruction of the lungs. Why some people get it and others not, is a mystery. It causes weight loss; fatigue,

breathlessness and sufferers cough up blood from the damaged lung tissue. As the infection progresses people are unable to work, become dependant on their families and will eventually die (80 per cent over five years). Antifungal therapy alleviates symptoms and prevents progressive lung destruction in about 60 per cent. CPA is often diagnosed late and only the underlying lung problem is recognised so treatment is often started at a much more advanced stage when quality of life is unrestorable."

Doug's problems started in 1985 when he was diagnosed with TB. For years he was treated for the condition but it was not until ten year's later that he was transferred to Wythenshawe Hospital where doctors told him he had developed CPA. He said: "To be honest I had never heard of aspergillosis but with a change in treatment I started to feel a bit better. I've had my setbacks. Five years ago I developed a resistance to oral drugs and my wife Chris had to learn how to give me my medication through a drip. I'm not complaining. This is the best I've felt in ages."

The National Aspergillosis Centre, based at University Hospital of South Manchester (UHSM)'s Wythenshawe Hospital is the world's leading unit for CPA with over 300 patients under treatment. Their prior research findings have identified several defects among 168 known immunity gene variants – this study extends to all human genes to discover completely new genetic variants.

Professor Ian Jacobs, Vice President of The University of Manchester and Director of MAHSC (Manchester Academic Health Science Centre) believes this pioneering study has the potential to deliver new diagnostic tests and treatments for patients both in the UK and across the world. He said: "The key to better treatment of CPA lies in unravelling the enormously complex risk profile of patients with the disease. Lung defects plus specific genetic variants are the root cause, and once *Aspergillus* gets into the lung in CPA, it is incurable."

Other team members include Dr Paul Bowyer and Dr Nicola Smith of the Manchester Fungal Infection Group, based at The University of Manchester, and Professor Angela Simpson of the NIHR Clinical Research Facility at the hospital.

Provided by University of Manchester

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