Scientist makes major cystic fibrosis breakthrough

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A Queen's University doctor has played a key role in a major breakthrough to change the lives of cystic fibrosis sufferers.

Queen's University's Professor Stuart Elborn, an international authority in respiratory medicine, with colleagues from the United States and Australia have led pivotal studies of a new treatment for people with Cystic Fibrosis. The combination therapy, developed by Vertex (a Boston, USA company), improves lung function and reduces hospitalisations for patients with the most common type of cystic fibrosis.

Two Phase 3 studies of the drugs ivacaftor and lumacaftor, which included over 1,100 patients worldwide, built on previous studies of ivacaftor in patients with G551D and other related mutations. Ivacaftor is the first drug to treat the underlying causes of cystic fibrosis rather than just its symptoms and is currently approved for patients with the 'celtic gene' mutation carried by about four per cent of all patients and 10-15 per cent of patients in Ireland. This therapy is a leading example of precision medicine, where treatment is based on a test for genetic mutations.

This recent trial looked at the treatment of patients with two copies of the F508DEL mutation which is carried by roughly half of all cystic-fibrosis patients. It found that a combination of ivacaftor with lumicaftor was effective in improving lung function between 2.6 and 4 per cent

Cystic fibrosis is a fatal lung disease affecting 75,000 children and adults world-wide, and is caused by inherited genetic mutations that vary among different patient groups.

Professor Stuart Elborn, Dean of the School of Medicine, Dentistry and Biomedical Sciences at Queen's University, and the European lead on the study, said: "This is a very significant breakthrough for people with cystic fibrosis. While we had previously found an effective treatment for those with the 'celtic gene' this new combination treatment has the potential to help roughly half of those with cystic fibrosis, those who have two copies the F508DEL mutation.

"This is another example of how Queen's scientists are working internationally to change lives around the world."

Provided by Queen's University Belfast