Cystic fibrosis drug reverses genetic abnormality in the CF mutation

November 28 2011, by Marion Lopez

The Lung Institute of WA (LIWA) has recently made a breakthrough in the search for a drug to improve the quality of life of patients with Cystic Fibrosis (CF).

A debilitating disease, CF is an inherited defect in a gene called CFTR (or Cystic Fibrosis Transmembrane Conductance Regulator) that codes for a complex protein involved in moving salt (Na and Cl) at cell surfaces.

According to LIWA's Director Professor Phil Thompson, the gene anomaly leads to defective mucus in the airways (stickier) and also in the pancreas affecting digestion of food and sweat glands.

"In the lung, this leads to recurrent infections, airway wall damage as well as inflammation, and leads to chronic lung disease resulting in early death," he says.

"The gene has many mutations and this leads to the various names such as G551D and F508del.

"Some mutations cause greater severity of abnormality than others."

The new drug treatment (VX-770), whose study results have been published in the New England Journal of Medicine, works by reversing the genetic abnormality in the CF mutation G551D.

The results showed a significant and sustained improvement in lung function, a lower rate in pulmonary exacerbations and a reduction in sweat chloride and weight gain.

Perth man Ernie Bindel was diagnosed with CF mutation G551D and has been on the trialed drug for over a year. He talks about the positive impact the medication has had on his condition.

"This is the longest period I have remained out of hospital in my life," he says.

"Over all I feel better, cough less and can take part in more physical activities.

"Another positive is that no needles were involved and the orally administered drug was quicker and easier to take than the intravenous injection."

LIWA's positive results encouraged the search for a drug treatment for F508del, the most common CF mutation which affects about 300 CF patients in WA.

LIWA is currently recruiting patients with the mutation to assess the drug VX-809, hoping the results will be similar to those experienced with mutation G551D.

Provided by ScienceNetwork Western Australia