First step towards treating rare childhood disease

July 17 2014

(Medical Xpress)—Scientists at Newcastle University have discovered a way of treating a rare disease which leads to kidney failure and death in children.

Although there is currently no cure for Joubert Syndrome, the new findings, published in the journal *Proceedings of the National Academy of Sciences*, mean that it is now possible to develop a therapy to help those suffering with the condition.

Joubert syndrome is an inherited developmental disorder affecting the brain, kidneys and eyes. Affected children have a range of problems including learning difficulties, movement problems, loss of eye sight and life-threatening cystic kidney disease, often leading to total kidney failure by the age of 13. A child suffering from this disorder can expect to spend up to 12 hours a day on dialysis, having a huge impact on their quality of life.

Over 4,000 people across the UK require renal replacement therapy (dialysis and transplantation) due to cystic kidney disease.

Publishing their findings this week, Dr John Sayer, a kidney specialist at Newcastle University and the Freeman Hospital, and Dr Colin Miles, a geneticist at the Institute of Genetic Medicine, Newcastle University, describe how they have discovered a cell signalling problem which causes cystic kidney disease in Joubert Syndrome.

"What is crucial here is that we have shown that the kidney damage in these patients is not permanent and so can potentially be treated," explains Dr Sayer.

"In Joubert syndrome, the kidney cells form abnormal cysts, meaning they are incapable of carrying out their role properly. We have found this happens because a defect in the cell's antenna, called the cilium, means the kidney cells can't communicate with each other properly. Using a new treatment we can restore this defect."

Several years of research has culminated in the discovery of a drug that can correct these damaged kidney cells from a patient in the laboratory.

Whist further research and testing is still needed, the work of Drs Sayer and Miles demonstrate that the abnormal cells in patients with cystic kidney disease are not permanently disabled and do respond positively to drug treatments. It also allows for personalised medicine approaches as treatments can be tested using cells obtained from a patient's urine sample, which are grown in the lab to form mini kidney tubules.

Dr Sayer adds: "Joubert syndrome is a dreadful condition that blights the lives of those who suffer from it. Hopefully this key breakthrough will provide the basis on which we can start to develop specific treatments."

Provided by Newcastle University