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Research by South Tyneside and Sunderland kidney expert brings hope to patients with inherited kidney diseases

A leading UK kidney consultant working with South Tyneside and City Hospitals Sunderland NHS Foundation Trusts has been instrumental in a breakthrough by Newcastle University researchers which is paving the way for genetic therapies for inherited kidney diseases.

Dr Shalabh Srivastava and fellow researchers at Newcastle University's Institute of Genetic Medicine, led by Professor John Sayer, identified for the first time how to

halt kidney disease in Joubert syndrome, a brain disorder causing varying degrees of physical, mental and, sometimes, visual impairments.

Patients with Joubert syndrome who also carry the *CEP290* gene develop cystic kidney disease, which leads to kidney failure and may require a transplant or dialysis. The researchers found it was possible to use a strand of engineered DNA to trick the cells' own editing machinery to bypass the *CEP290* mutation that causes kidney damage – a technique known as 'exon-skipping'.

Using this technology, they were able to dramatically improve kidney disease in a mouse, opening up the possibility of editing out genetic mistakes that are leading to inherited kidney diseases such as Joubert syndrome. The technology is being tested further before moving on to patient studies. It is expected that testing treatment of patients with exon-skipping will begin within the next three years.

A paper about the scientific breakthrough, of which Dr Srivastava is co-lead author, has now been published in one of the world's leading scientific journals, PNAS - Proceedings of the National Academy of Sciences of the United States of America.

Dr Srivastava said: "Gene editing to the kidney had previously been thought to be too difficult so it is very exciting to be part of such a significant development which has the potential to help patients with life-limiting, inherited kidney conditions all over the world by offering genetic therapy to correct the gene mistake within kidney cells."

Professor John Sayer, from the Institute of Genetic Medicine, Newcastle University, said: "This research paves the way for many genetic diseases of the kidney to be treated by gene editing.

"This regional collaboration between Newcastle University and local Trusts allows us to identify patients with inherited kidney disease and involve them in the latest research."

Since joining South Tyneside and City Hospitals Sunderland NHS Foundation Trusts in 2017, Dr Srivastava has introduced a dedicated Inherited Kidney Disease and Stone Clinic for patients throughout the North East and North Cumbria. The only other such clinic in the region is at Freeman Hospital, Newcastle.

Dr Srivastava added: “This new clinic, which was only made possible due to the joint working between our two Trusts, brings the most cutting edge diagnostic and therapeutic interventions in inherited kidney conditions closer to patients.”

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Photo caption: From the left, Dr Shalabh Srivastava and Professor John Sayer with other members of the research team at Newcastle University'

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