Research describes the diversity of genetic changes that cause inherited kidney disease in Ireland

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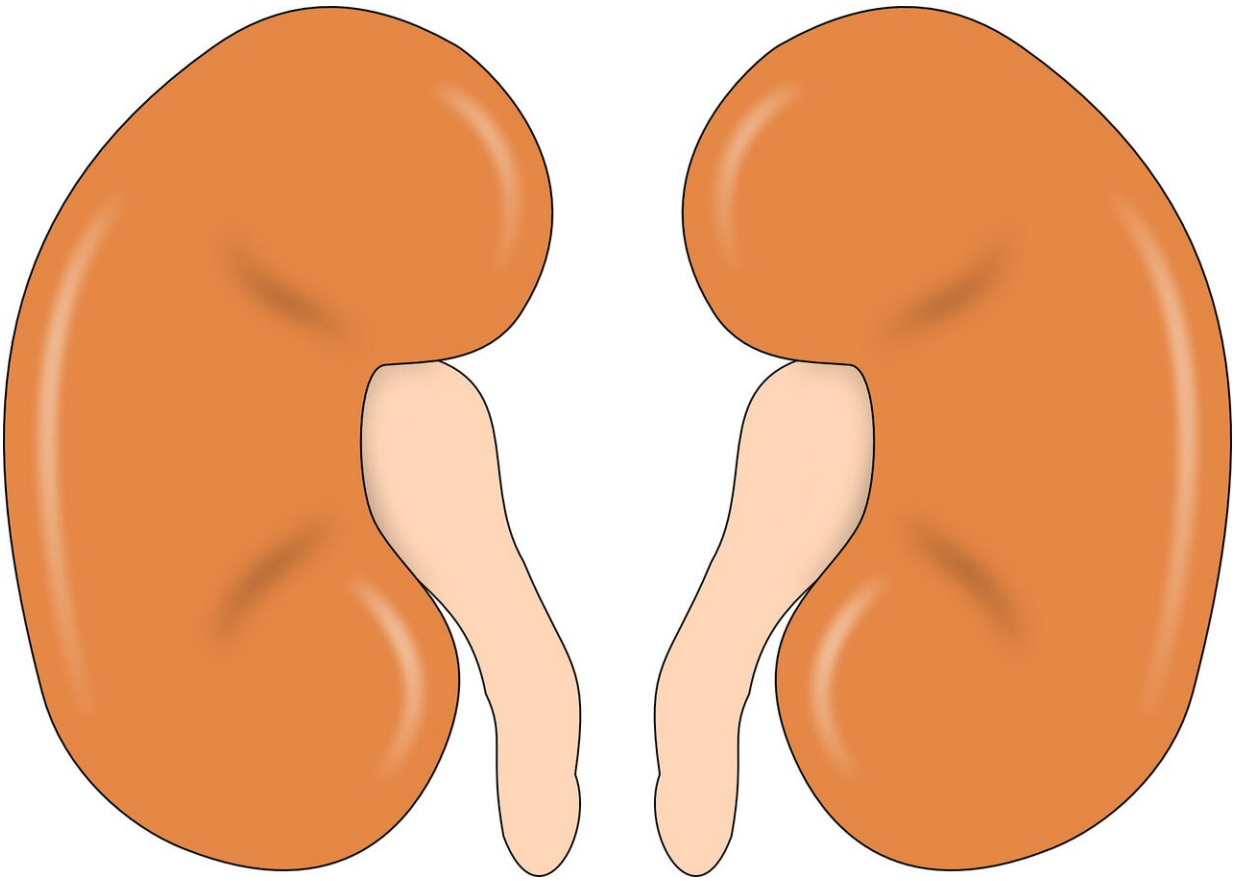

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A study has described genetic changes in patients with the most common

form of hereditary kidney disease that affects an estimated 12.5 million people worldwide. The research, which focussed on Polycystic Kidney Disease (PKD) in Ireland, provides insights into PKD that will assist doctors and patients in the management of this of inherited condition.

The study, led by researchers from the RCSI University of Medicine and Health Sciences, is published in the *European Journal of Human Genetics*.

In the research, a cohort of 169 patients with PKD in Ireland were analysed. The genetic changes were identified in up to 83% of cases. It is the first time that the diversity of genetic causes of PKD in Ireland have been described. The results will better assist doctors in identifying patients who may require transplantation or dialysis. The findings also have important implications for people who have a family history of PKD and are planning a family or considering kidney donation.

"This study is hugely important in providing us with an insight into the genetic landscape of Polycystic Kidney Disease, the most common form of inherited kidney disease in the world," said first author on the study Dr. Katherine Benson, School of Pharmacy and Biomolecular Sciences, RCSI.

"Our findings have implications for the prognosis of patients by helping us to further identify why the disease may progress more rapidly in some cases and how we can reduce the burden of inherited kidney disease in future."

More information: Katherine A. Benson et al. The genetic landscape of polycystic kidney disease in Ireland, *European Journal of Human Genetics* (2021). DOI: 10.1038/s41431-020-00806-5