



Purespring announces publication of notable study highlighting an under recognised genetic driver of adult-onset FSGS, in particular in US and UK populations

- Population modelling study published in *Kidney International Reports*
- Analysis of over 1.2 million genomes from five international databases predicts a substantial, previously underdiagnosed adult NPHS2-FSGS population
- Approximately 77% of cases in the US are predicted to involve two different NPHS2 variants (compound heterozygosity), most commonly R229Q paired with A284V. A284V is particularly prevalent in mixed-ancestry populations
- This modelling is supported by real world cohorts taken from clinical trial and gene panel data
- Finding emphasises the importance of genetic screening in adult FSGS, especially in patients resistant to immunosuppression, to establish the cause of FSGS and thereby the ideal therapy

London – 29 April 2026 – Purespring Therapeutics (Purespring or the Company), a precision nephrology company advancing targeted genetic therapies to preserve kidney function in underserved populations, today announced the publication of new research demonstrating that pathogenic NPHS2 variants are a significant and under-recognised cause of adult-onset focal segmental glomerulosclerosis (FSGS).

The study, *'Population modeling defines adult-onset focal segmental glomerulosclerosis caused by NPHS2 pathogenic variants'* published in *Kidney International Reports*, was conducted in collaboration between Purespring Therapeutics, Bristol Renal at the University of Bristol, Genescape, Natera and Travers Therapeutics. Pathogenic variants in the NPHS2 gene, which encodes podocin – a critical component of the kidney's filtration barrier – represent one of the most common causes of genetic FSGS, particularly in children. Paediatric-onset FSGS is relatively well recognised, and often associated in Northern European and US populations with the pathogenic NPHS2 variant R138Q.

In contrast, other NPHS2 variants are less well studied and may only result in disease when inherited in trans, that is, on the opposite copy of the gene from a second pathogenic variant. One such variant is R229Q, which occurs at relatively high frequency in UK and European populations but is typically non-pathogenic in isolation. However, particularly in the North American admixed population, R229Q may be co-inherited with other pathogenic variants – most notably A284V – leading to adult-onset FSGS.

The study combines population modelling of more than 1.2 million genomes from five international databases with validation in real-world cohorts. The findings suggest a substantial adult patient population across the US, driven largely by R229Q compound heterozygosity, predicted to account for approximately 77% of US NPHS2-driven kidney disease and frequently occurring alongside the A284V variant in individuals with admixed Latin American ancestry.

The data suggest that NPHS2-associated nephropathy is more common than previously recognised and may present as slowly progressing nephrotic syndrome in which patients reach end stage renal disease (ESRD) in adulthood rather than in childhood. Genetic screening of adult patients with FSGS, particularly those who are immunosuppression-resistant, may help to identify underlying monogenic disease and guide more appropriate, targeted treatment strategies. These results are supported by clinical cohort data, including analyses from Natera's RenaSight™ platform and Trave's DUPLEX study, where the majority of adult patients with NPHS2-associated disease carried R229Q variants.

To read the full paper in *Kidney International Reports*: [Population modelling defines adult-onset focal segmental glomerulosclerosis caused by NPHS2 pathogenic variants](#)

Fredrik Erlandsson, Chief Medical Officer at Purespring Therapeutics commented: "This study reinforces our leadership in advancing the understanding of genetic kidney disease and its application in precision nephrology, and the strength of our highly collaborative approach. Our findings highlight a substantial, previously undiagnosed population of adults with NPHS2-mediated FSGS. This underscores a clear need to expand genetic testing, improve diagnosis and enable more targeted treatment strategies for patients."

Moin Saleem, Professor of Paediatric Renal Medicine at University of Bristol and Founder and Chief Scientific Advisor at Purespring Therapeutics said: "We are delighted to have worked with Purespring, Trave, Natera and Genescape to identify and quantify the genetic basis behind NPHS2-driven nephropathy, an important and under-recognised sub-set of FSGS patients. The results reinforce the importance of integrating genetic screening across paediatric and adult renal care, not only for diagnosis but also for guiding treatment decisions, and we hope they lead to increased uptake in clinical practice."

The study will help inform the development strategy and regulatory pathway for PS-001, Purespring's candidate currently in development as a novel treatment for FSGS caused by pathogenic NPHS2 variants.

For further information, contact:

Purespring:

contact@purespringtx.com

+44 (0)20 3855 6324

[LinkedIn](#)

ICR Healthcare:

Sarah Elton-Farr, Emily Johnson

purespring@icrhealthcare.com

About Purespring

Purespring Therapeutics, a precision nephrology company pioneering podocyte-targeted, genetically-driven therapies to preserve kidney function where unmet need is greatest. Through its proprietary

GlomThera™ platform, Purespring is able to deliver genetic therapies directly to the podocyte, offering a novel approach to the treatment of kidney diseases.

Purespring's multi-asset pipeline targets multiple renal indications with significant unmet medical need. The Company's lead programme, PS-002, offers a highly differentiated approach for patients with IgA nephropathy (IgAN). By precisely targeting the site of disease, Purespring aims to transform the trajectory of kidney disease so patients can live fuller, healthier lives.

Purespring is backed by leading biotech investors, including Syncona Limited, Sofinnova Partners, Gilde Healthcare, Forbion, and the British Business Bank and has raised £115m (\$150m) to date.

For more information please visit: purespringtx.com and follow us on [LinkedIn](#).