

WASHINGTON UNIVERSITY PATHOLOGY SERVICES

RENAL DISEASE TESTING

Genetics and Genomic Services offers renal disease panels designed in collaboration with Washington University School of Medicine Nephrologists and Renal Subspecialty Pathologists. Results provide physicians with definitive information to make accurate, timely diagnoses. Testing spans routine patient care and clinical translational research, allowing for rapid translation of new gene- and variant-diseases relationships.

EXPERT CONSULTATION AND DIAGNOSIS

- Next-generation sequencing (NGS) for efficient, cost-effective testing designed to arrive at timely and accurate diagnoses
- Concise, expert interpretations by experienced faculty board-certified in clinical genomics
- Collaborative consultations with renal subspecialty pathologists and nephrologists with expertise in renal genetics are available as needed

COMPREHENSIVE DIAGNOSTICS

- Exome capture NGS for comprehensive and deep coverage of all coding exons of ordered genes with robust detection of single nucleotide variants (SNVs) and small insertions and deletions (indels)
- Discrete orderable gene panels organized according to clinical presentations consistent with nephrotic syndrome and focal segmental glomerulosclerosis, complement-mediated renal disease, cystic diseases and nephronophthisis, and Alport Syndrome
- Exome-level data for reanalysis on indeterminate cases, or those with negative findings, as indicated
- Send out testing via MLPA for detection of large deletions in the *CFHR3-CFHR1* gene cluster for the Complement-Mediated Renal Disease panel
- Reflex send-out testing by aCGH for large deletion/duplication in other loci, as clinically appropriate

FOR MORE INFORMATION OR TO ORDER A TEST, CONTACT:

Phone: 314-747-7337

Email: gps@wustl.edu

Web: pathologyservices.wustl.edu



SPECIMENS ACCEPTED

- 2 to 5 mL peripheral blood in lavender top EDTA tube
- 2 to 4 buccal swabs
- Specimen kits available upon request

RENAL TEST MENU

- Alport Syndrome Panel (3 genes)
- Complement-Mediated Renal Disease Panel (13 genes)
- Cystic disease and Nephronophthisis Panel (32 genes)
- Nephrotic Syndrome and Focal Segmental Glomerulosclerosis Panel (52 genes)

STREAMLINED ORDERING, REPORTING AND BILLING

- Four weeks average turnaround time
- Reporting can be done via fax or electronically
- Assistance with the insurance prior authorization process and determination of patient out of pocket costs for most insurances



Pathology & Immunology