For individuals affected by kidney disease, genetic testing can identify underlying heritable causes, enable diagnosis, and provide insights on prognosis, clinical management, and risk of recurrence. Genetic testing can also help providers understand the need for additional screening recommendations for patients and family members.

Since more than 10% of kidney disease cases in the United States are thought to be genetic in origin, genetic testing is suggested for all patients with chronic kidney disease.

**Advanced analysis to diagnose genetic disease**

Our renal genetic testing was developed by a team of Mayo Clinic nephrology and genetic experts and is among the most comprehensive diagnostic testing available. Our genetic panels use next-generation sequencing to evaluate clinically relevant genes across a number of disease states. In addition, these evaluations:

- Provide the maximum depth of coverage, highest possible detection rates, and low false negative and false positive rates across all included genes.
- Detect both small variants, including single nucleotide variants and insertions and deletions, and copy number variants.
- Use long-range polymerase chain reaction (PCR) analysis to detect variants in technically challenging genes, such as PKD1.
- Provide standard coverage of +/-10 base pairs flanking each coding exon along with expanded coverage to detect rare, clinically actionable, intronic genetic variants.
- Classify variants based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance.
- Are validated to detect 95% of deletions up to 75 base pairs (bp) and insertions up to 47 base pairs.
COMPREHENSIVE EVALUATION FOR KIDNEY DISEASE

Key testing

NEPHP | Comprehensive Nephrology Gene Panel, Varies

For individuals affected by kidney disease, genetic testing can identify underlying heritable causes, enable diagnosis, and provide insights on prognosis, clinical management, and risk of recurrence. In addition, genetic testing can help providers understand the need for additional screening recommendations for patients and family members.

This test uses next-generation sequencing to evaluate 302 clinically relevant genes across a number of disease states.

Advantages

• Provides a genetic evaluation for patients with a personal or family history suggestive of hereditary kidney disease.

• Can establish a diagnosis for a variety of hereditary kidney conditions including focal segmental glomerulosclerosis, nephritic/nephrotic syndrome, Alport syndrome, cystic kidney diseases (including polycystic kidney disease), and nephronophthisis.

DISEASE-FOCUSED PANELS

Key testing

AHUGP | Atypical Hemolytic Uremic Syndrome (aHUS)/Thrombotic Microangiopathy (TMA) Complement 3 Glomerulopathy (C3G) Panel, Varies

This panel evaluates for aHUS, TMA, and C3G, conditions which cause individuals to display complement activation of the microvasculature, abnormalities in the walls of small blood vessels, dense deposit disease, or C3 glomerulonephritis.

Advantages

• Provides a genetic evaluation for patients with a personal or family history suggestive of aHUS, TMA, or C3G.

• Establishes a diagnosis of genetic aHUS, TMA, or C3G and, in some cases, allows for appropriate management and surveillance for disease features based on the gene involved.

• Identifies variants in genes encoding complement alternate pathway components and specific coagulation pathway genes known to be associated with increased risk for aHUS, TMA, and C3G, allowing for predictive testing of at-risk family members.

ALAGP | Alagille Syndrome Gene Panel, Varies

Approximately 40% of individuals with Alagille syndrome (ALGS) also have kidney disease. The kidney features may include structural abnormalities like small hyperechoic kidney, kidney cysts, or ureteropelvic obstruction as well as kidney dysfunction, such as renal tubular acidosis. This test assesses two genes associated with Alagille syndrome.

Advantages

• Uses next-generation sequencing to detect single nucleotide, small deletion-insertion, and copy number variants in two genes (JAG1, NOTCH2) associated with Alagille syndrome.

• Provides a genetic evaluation for patients with a personal or family history suggestive of Alagille syndrome.

• Establishes a diagnosis of Alagille syndrome.

ALPGP | Alport Syndrome Gene Panel, Varies

This panel evaluates for Alport syndrome (AS), a genetic disorder caused by variants in genes that are involved in the formation of a basement membrane collagen network. AS is characterized by kidney disease, sensorineural hearing loss, and ocular findings, and, in many cases, leads to kidney failure.

Advantages

• Uses next-generation sequencing to identify disease-causing variants in four genes (COL4A3, COL4A4, COL4A5, and COL4A6) known to be associated with Alport syndrome to help with patient diagnosis and management.

CASRG | CASR Full Gene Sequencing with Deletion/Duplication, Varies

The CASR gene is associated with several disorders of calcium regulation:

• Autosomal dominant familial hypocalciuric hypercalcemia (FHH)

• Autosomal dominant primary hyperparathyroidism (PHPT)

• Autosomal dominant and autosomal recessive neonatal severe hyperparathyroidism (NSHPT)

• Autosomal dominant hypocalcemia (also known as hypoparathyroidism)

• Autosomal dominant hypocalcemia with features of Bartter syndrome

Advantages

• Uses next-generation sequencing to identify disease-causing variants associated with CASR-related disorders.

• Includes analysis of CASR and can assist with diagnosis, prognosis, and clinical management of patients with CASR-related disorders.

RBART | Bartter Syndrome Gene Panel, Varies

Bartter syndrome is a rare, hereditary tubulopathy of highly variable severity that can cause renal salt-wasting in infants and young children due to impaired sodium/chloride reabsorption in the thick ascending limb of the nephron.

Advantages

• Uses next-generation sequencing to detect single nucleotide, deletion-insertion, and copy number variants in six genes associated with Bartter syndrome: BSND, CLCNKA, CLCNKB, KCNJ1, MAGED2, and SLC12A1.

• Establishes a diagnosis of Bartter syndrome.

• Provides a genetic evaluation for patients with a personal or family history suggestive of Bartter syndrome.
**CYSTIC KIDNEY DISEASE**

### Key testing

- **RSCGP | Nephrocalcinosis, Nephrolithiasis, and Renal Electrolyte Imbalance Gene Panel, Varies**

Hereditary forms of cystic kidney disease have several underlying etiologies and may present in childhood or adulthood, with or without extrarenal features. The two most common categories of hereditary cystic kidney disease are ciliopathic disorders and phakomatoses. Ciliopathic disorders causing cystic kidney disease include polycystic kidney disease (PKD), nephronophthisis (NPHP), and medullary cystic kidney disease (MCKD).

### Advantages

- Uses next-generation sequencing to identify disease-causing variants in 45 genes associated with cystic kidney disease.
- Detection of a disease-causing variant may assist with diagnosis, prognosis, clinical management, familial screening, and genetic counseling for cystic kidney disease.
- Provides a genetic evaluation for patients with a personal or family history suggestive of hereditary cystic kidney disease.

### HEREDITARY CAUSES OF KIDNEY STONES

### Key testing

- **RFSGS | Renal Stone Gene Panel, Varies**

When the presence or severity of electrolyte imbalance or kidney stones observed in a patient cannot be explained by acquired causes or there are multiple cases clustered within a family, genetic testing for the inherited causes of kidney or extrarenal impairment of osmoregulation may be considered.

### Advantages

- Assesses 72 genes associated with nephrocalcinosis, nephrolithiasis, and other inherited conditions associated with renal electrolyte imbalances.
- Establishes a diagnosis for a variety of hereditary conditions associated with renal salt wasting or abnormal salt retention, impaired acid-base, water, and calcium homeostasis, or kidney crystallization.

### HEREDITARY CAUSES OF FSGS AND NEPHROTIC SYNDROME

### Key testing

- **APOL1 | APOL1 Genotype, Varies**

Nephrotic syndrome (NS) is a kidney disorder characterized by proteinuria, hypalbuminemia, and edema. Many conditions can cause NS, including diseases that only affect the kidney and other more systemic disorders, such as diabetes or lupus. FSGS, a histologic finding characterized by sclerosis involving part of the kidney glomeruli, is commonly found in patients with NS.

### Advantages

- Uses next-generation sequencing to detect single nucleotide, deletion-insertion, and copy number variants in 56 genes associated with focal segmental glomerulosclerosis and NS. This test also includes targeted assessment and reporting of the G1 and G2 alleles of the APOL1 gene.
- Provides a genetic evaluation for patients with a personal or family history of FSGS and/or steroid resistant nephrotic syndrome (SRNS).
- Establishes a diagnosis of inherited form of FSGS and/or SRNS and guides treatment decisions in individuals with FSGS and/or NS.

### CUSTOMIZED NEPHROLOGY GENE PANELS

### Key testing

- **CGPH | Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies**

This panel is customizable, allowing for the inclusion of desired genes.

### Advantages

- Detects single nucleotide and copy number variants in a custom gene panel.
- May identify a pathogenic variant that can assist with diagnosis, prognosis, clinical management, familial screening, and genetic counseling for a hereditary condition.
GENETIC KIDNEY DISEASE


FOR CLINICAL OR TECHNICAL SUPPORT CONTACT OUR SPECIALISTS:
855-516-8404 | +1-855-379-3115 (INTERNATIONAL)
NEWS.MAYOCLINICLABS.COM/RENAL

©2023 Mayo Foundation for Medical Education and Research. All rights reserved. MAYO, MAYO CLINIC, Mayo Clinic Laboratories, and the triple-shield Mayo logo are trademarks and service marks of MFMER.