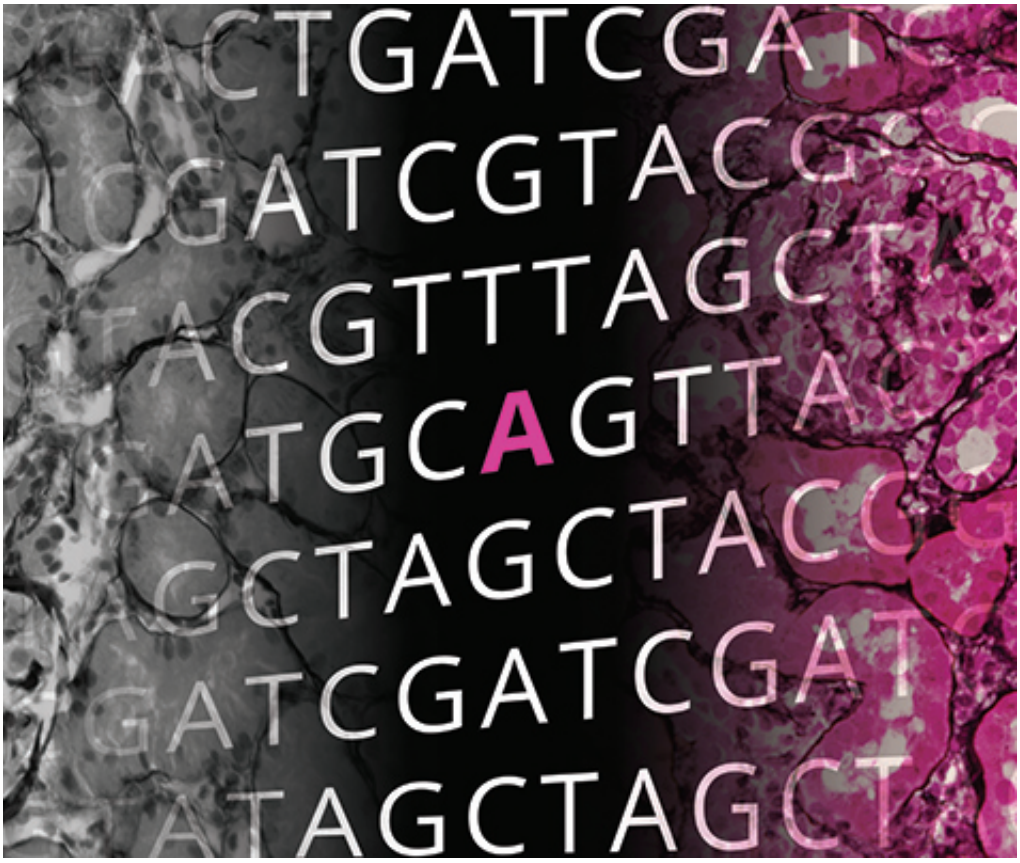
Genetic test for inherited kidney diseases developed

March 27 2015, by Michael C. Purdy And Gaia Remerowski



A new test from Washington University's Genomic Pathology Services will help physicians quickly zero in on genetic mutations that may be contributing to kidney disease. Credit: GPS

Many kidney disorders are difficult to diagnose. To address this problem, scientists and clinicians have developed a diagnostic test that

identifies genetic changes linked to inherited kidney disorders. This testing is now available nationwide through Genomic Pathology Services (GPS) at Washington University School of Medicine in St. Louis.

"For many kidney diseases, diagnosis can be an odyssey in which you sequence one gene after another over a long period of time to learn what's going wrong and what the best options are for treatment," said GPS chief medical officer and Washington University pathologist Jonathan Heusel, MD, PhD. "It makes more sense to screen all the possible contributing genes at once with a single test and consider options for treatment."

To make this possible, the GPS team developed the test with kidney disease specialists, including Joseph Gaut, MD, PhD, a renal pathologist.

The test employs next-generation sequencing technology to decode genes associated with kidney disease. Using software developed at the university, clinical genomics specialists analyze and interpret the observed genetic alterations to identify disease-related genetic changes, or variants. They then must determine whether a given variant poses clinical risks based on available medical knowledge.

"The variants have to be evaluated on a case-by-case basis, which can be time-consuming and labor-intensive," Heusel said.

GPS continues to update the kidney test as new links between kidney problems and DNA are identified.

"We stay abreast of the literature, and as new genes become clinically meaningful, we will incorporate those into the test," said Catherine Cottrell, PhD, medical director for GPS.

The kidney test will check for:

- Alport syndrome, which is characterized by progressive loss of kidney function, hearing loss and eye abnormalities;
- Nephrotic syndrome, which includes symptoms such as protein in the urine, low blood-protein levels, high levels of cholesterol and triglycerides, and swelling;
- Metabolic disorders associated with renal disease and including other systemic abnormalities such as diabetes, amyloidosis and others;
- Complement (immune system) defects related to kidney disease, including atypical hemolytic uremic syndrome.

Physicians interested in kidney genetic testing for their patients may contact GPS, which assists with insurance preauthorization for the service.

For the analysis, physicians submit a blood sample from the patient. GPS analyzes the sample and sends the physician a report outlining the results in four to six weeks.

GPS recommends that physicians make genetic counseling available to patients to help them understand and manage their diagnoses and any health implications for family members.

More information: For more information, contact GPS by phone at 314-747-7337 (toll-free 866-450-7697), email at gps@wustl.edu or visit the website: gps.wustl.edu/

Provided by Washington University School of Medicine in St. Louis