# Gene testing for heart diseases now available

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A mutation, highlighted with a red letter in the genetic code shown above, can alter the heartbeat in dangerous ways. A new panel offered by Genomic Pathology Services can test up to 69 genes at once for changes that affect heart function.

Washington University School of Medicine in St. Louis now offers genetic testing to help diagnose and treat patients with heart disorders that can lead to sudden death.

The new test, offered though the school's Genomics and Pathology Services (GPS) and developed in collaboration with Washington University cardiologists, analyzes genes linked to arrhythmias and cardiomyopathies. Physicians who treat patients with indications of these heart conditions can submit a blood sample for gene sequencing. In two

to three weeks, they receive a report from GPS describing any mutations identified in the patient's DNA that may contribute to disease and affect response to treatment.

"Results from this test can help us refine diagnoses and allow us to personalize management and treatment of our patients," said Phillip Cuculich, MD, assistant professor of medicine and one of the cardiologists involved in the development of the test. "The results also may help family members of affected patients decide if they want to be tested to see if they are at risk for developing the disease."

Called the Washington University CardioGene Set, the new test builds on another first-of-its-kind test offered by GPS that analyzes multiple genes in tumors. Seeking genomic clues to personalize cancer diagnosis and treatment, oncologists nationwide have been sending tissue samples to GPS for this test for two years. The cancer gene set recently expanded from 25 genes to 40 genes.

"The CardioGene Set is the next major step in our effort to bring the promise of human genomics to the clinic," said Karen Seibert, PhD, director of GPS. "With rapid and sensitive gene-sequencing technology, we check dozens of heart disease genes simultaneously to cost-effectively identify the likely genetic cause."

The cardiac testing panel includes genes linked to eight cardiac disorders that are characterized by irregular heartbeats (arrhythmias) or heart muscle problems (cardiomyopathies). Testing is traditionally offered one gene at a time, but the new test can report on as many as 69 genes, saving money and time.

For example, the test analyzes the sequences of genes linked to long-QT syndrome, a rare inherited arrhythmia that lengthens the time between heartbeats, potentially causing heart palpitations or cardiac arrest. These

genes can help doctors identify particular subtypes of long QT. Different forms of long QT respond to different therapies, so this information helps guide treatment decisions.

Patients with hypertrophic cardiomyopathy (HCM), a thickening of the heart muscle that can lead to sudden cardiac arrest and other problems, also can benefit from the CardioGene Set. HCM is infamous for causing sudden fatal cardiac arrests in young athletes who never showed any symptoms of heart problems. Genetic diagnosis of this condition may lead physicians to advise a patient against physical overexertion.

Work is underway to develop several additional genetic testing panels, including panels focused on autism genes and kidney disease genes.

Provided by Washington University School of Medicine in St. Louis