PUTTING PRECISION HEALTH INTO PRACTICE

STANFORD MEDICINE'S CLINICAL GENOMICS SERVICE IS LEADING HEALTH CARE INTO A NEW ERA
TO ACHIEVE THE LIFE-CHANGING POSSIBILITIES of Precision Health, we first have to deliver the fundamentals of precision medicine—including genomic sequencing that is accurate, fast, and affordable. A pilot program in clinical genomics is allowing a small group of patients at Stanford Health Care and Lucile Packard Children’s Hospital Stanford to have their DNA deciphered to help doctors with diagnosis and treatment. The goal of the Clinical Genomics Service is to help doctors better diagnose and treat genetic disorders. In the pilot phase, genomic testing will be limited to patients with “mystery” diseases (typically children), patients with unexplained hereditary cancer risk, patients with inherited cardiovascular or neurological disease, and those with severe, unexplained drug reactions. Potential participants must first be referred by a physician and the Clinical Genomics team will then determine if cases are suitable for sequencing.

“I’m very excited to bring the pioneering work of Stanford genomic scientists to the bedside of our patients,” said Euan Ashley, MRCP, DPhil, associate professor of medicine and of genetics and co-director of the Clinical Genomics Service. “Thanks to the foresight and support of our leadership, we have the opportunity to bring world-leading Stanford science to Stanford patients fast and first.”

Over the past few years, genome sequencing has demonstrated great promise, particularly in cases of rare diseases, where patients and families may have a genetic condition that has eluded diagnosis. Already, hundreds of families with genetic diseases have benefited from the power of this technology to discover the underlying causes of their conditions. In some cases, identifying a genetic cause of disease may also provide important information related to personalized therapy and care management. Stanford’s service uses a highly integrated approach that includes professional genetic counseling, the most advanced genome sequencing technology available, and expert interpretation by molecular genetic pathologists and other physicians with expertise in this emerging and complex field.

This service will be closely tied to other diagnostic genetic testing currently offered at the two hospitals. Those programs, which include molecular genetic pathology, cytogenetics, and clinical biochemical genetics, have an outstanding record of compliance with the extensive regulatory requirements for diagnostic genetic testing.

“Stanford has a special wealth of information and analysts,” said Jason Merker, MD, PhD, assistant professor of pathology, the service’s co-director. “We involved physicians, health care providers, bioethicists, bioinformaticians, and other researchers, inviting everyone to voice their thoughts for the broadest, deepest discussions possible on how to apply these new methods and knowledge to clinical care.”

Michael Snyder, PhD, the director of the Stanford Center for Genomics and Personalized Medicine (SCGPM), the Stanford W. Ascherman, MD, FACS, Professor in Genetics, and chair of the Department of Genetics, as well as members of the SCGPM, played a pivotal role in the foundation of the Clinical Genomics Service. Also included in those discussions were Carlos Bustamante, PhD, a Stanford professor of genetics who was named a 2010 MacArthur Fellow for his work in genetic sequencing, and Michael Cherry, PhD, Stanford professor of genetics and principal investigator in several genome database projects.

“This service can represent the best definition of the term personalized medicine,” said Amir Dan Rubin, president and CEO of Stanford Health Care. “The collaboration of our world-class experts in patient care and scientific research will advance the leading edge of knowledge in genome sequencing, bringing greater value, in the most responsible way, to what we offer our patients. Our goal is to use this new technology for early and accurate diagnosis and treatment for patients now—and to learn and share that knowledge with medicine’s new future.”

Lloyd B. Minor, MD, the dean of the School of Medicine, said the Clinical Genomics Service exemplifies the bench-to-bedside philosophy at the heart of Stanford Medicine. “It serves as a bridge to better connect the groundbreaking genetic science of our laboratories with the patient care of our hospitals and clinics. The efforts of Drs. Ashley and Merker and their team are helping to shape a medical future in which disease risk can be more accurately predicted and treatments better tailored to individual patients.”

Christopher G. Dawes, president and CEO of Lucile Packard Children’s Hospital Stanford and Stanford Children’s Health, sees the service as a logical extension of the kind of care established since the hospital’s opening—and its particular intellectual environment. “The genetics revolution is taking off in Silicon Valley, much like the information technology revolution has over the past few decades,” said Dawes. “We are very fortunate that our...we have a remarkable opportunity to bring world-leading Stanford science to Stanford patients fast and first.”

Stanford scientists work so closely with our pediatricians and obstetricians in conducting leading research in the field of genetics.

Stanford’s Clinical Genomics Service joins a small group of other medical centers that now offer a variety of sequencing options. In 2010, Ashley and Stephen Quake, PhD, the Lee Otteson Professor of Bioengineering, were the first in the world to use a healthy person’s complete genome sequence to predict risk for disease and anticipate reaction to several common medications. These new genomic services are the first wave to test this new knowledge. “As people who are in the trenches, I hope we can temper appropriate optimism with realism,” said Ashley.