Individualized Medicine

It's All in the Genes

Imagine being able to accurately predict your individual risk of developing certain diseases five, 10 or 20 years from now – and know how well your body will respond to various treatments. Though such highly individualized medicine sounds like a revolutionary concept, it's already happening at Scripps.

By studying the unique genetic structures and DNA that create a customized blueprint for every human being, Scripps researchers and physicians are more accurately and effectively diagnosing and treating illness, assessing future risk, and even helping prevent disease in individual patients.

During the past several years, Scripps has spearheaded a number of landmark studies designed to categorize and evaluate the genetic profiles of individual patients through state-of-the-art science and technology. From a first-of-its-kind genetic scan that identifies increased risk of a wide range of serious diseases to studies that help physicians tailor treatments according to specific gene variations, Scripps is at the forefront of individualized medicine research. Results of these groundbreaking genetic studies have already enabled Scripps physicians and researchers to offer individualized care to patients.

"Medicine today is widely practiced at a population level, with general guidelines for large groups," says Eric Topol, MD, director, Scripps Translational Science Institute. "At Scripps, we recognize that each individual is completely unique. Part of that can be determined through genomes, and part through physiological measurements like glucose, blood pressure, heart rhythm and other metrics. These enable us to establish far more information about individual patients than ever before, and translate that into valuable knowledge about their risks for disease, responses to treatment and prevention strategies."

Eric Topol, MD

"We can define a human being today in a way that has never been possible in the past, and that enables this whole concept of individualized medicine and true prevention, which is so exciting. This is a momentous time in medicine – to be able to understand an individual's unique 'black box.'"

– Eric Topol, MD, Director
Scripps Translational Science Institute

Determining Individual Genetic Risk

In 2007, Scripps was the first and only health system in the nation to offer an innovative genetic study that gave participants a detailed scan of their individual genome based on DNA. Scripps Genomic Health Initiative (SGHI) assessed individual genetic risk for more than 20 health conditions, including such diseases as breast cancer, colon cancer, glaucoma, heart attack, multiple sclerosis, Type 2 diabetes, Alzheimer's, lung cancer and macular degeneration.

Each participant received a personal genetic report, along with information and recommendations to help prevent various conditions through healthier behavior, such as diet and exercise. Participants agreed to track their lifestyle changes through follow-up studies over the next 20 years.

In January 2011, The New England Journal of Medicine published the first results from the study. More than a quarter of participants reported improving their diet and exercise, and about half said they planned to seek additional medical screenings, such as a mammogram or glucose test, as a result of their genetic profile.

"My mother and I enrolled in the Scripps Genomic Health Initiative together to see if we were genetically predisposed to any health conditions. As a result, we have both made lifestyle changes. I incorporate exercise in my daily routine, from taking the stairs at work to walking and riding my bike around the neighborhood."

– Sarah Clarke, Nurse Practitioner
Scripps Clinic Interventional Cardiology

Sarah Clarke, NP

Genetic Testing in Heart Care

Scripps was also first in the United States to apply genotyping to improve heart care. Already a renowned leader in cardiovascular care, Scripps now offers genetic testing to patients about to undergo stent procedures to determine their ability to respond to Plavix (clopidrogel), an anti-clotting drug given to most coronary stent patients. The test identifies common gene variants linked to the inability to metabolize Plavix; patients who have these variants carry double or triple the risk of death, heart attack or stroke compared to people without them. As a result of the testing and other metrics, physicians can offer these higher-risk patients treatment options and follow-up care tailored to their genetic profiles. Moreover, Scripps is moving forward with groundbreaking research to accelerate the genotyping process, enabling physicians to obtain results in less than an hour rather than several days.

"Thank you for helping me access the gene test. Though I had planned to switch to clopidrogel, the results indicated that I am a non-responder. I truly appreciate the wonderful work Scripps is doing in genomic medicine."

– Ed Funkhouser
Plavix Study Participant

Mathew Price, MD

Genetic testing for this population carries significant implications for patient care. More than 1 million people undergo stent procedures in the U.S. every year. The gene variants associated with an inability to metabolize Plavix occur in more than 30 percent of people of European ancestry and more than 40 percent of those of African or Asian ancestry.

In addition to genotyping, Scripps physicians use a bedside test to measure patients' platelet response to Plavix and determine how well the drug is working. Scripps also led an international multicenter randomized clinical trial, published in The Journal of the American Medical Association in March 2011, which studied the effectiveness of Plavix dosage changes based on patients' measured responses.

"We're not only using individualized medicine with our patients, we're moving the field forward," says Scripps Clinic interventional cardiologist Matthew Price, MD. "We know that people metabolize and respond to drugs differently and much of that is based on DNA. By using genetics and other novel diagnostic tests, we can select the right amount of the right drug for a particular patient, as well as measure the response to ensure the best results."

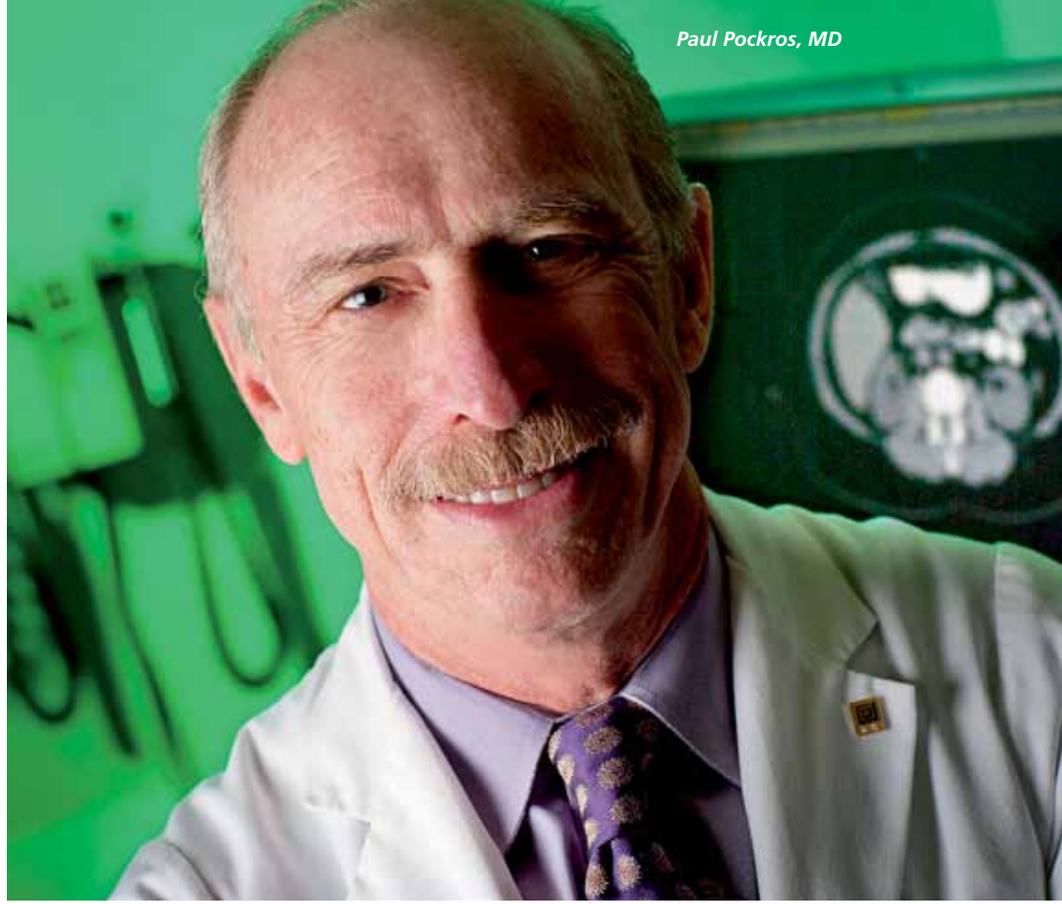
Personalizing Hepatitis C Treatment

Genetic testing under way at Scripps offers hope to the more than 4 million U.S. patients diagnosed annually with hepatitis C, many of whom suffer unpleasant side effects from interferon therapy. Commonly prescribed for this condition, interferon causes flu-like symptoms and costs more than $50,000 annually per patient. Scripps researchers use genetic testing to identify a common gene variant that predicts successful treatment using a combination therapy of interferon and ribavirin.

Specialists at Scripps Clinic now routinely order this genotyping on all hepatitis C patients who are potential candidates for antiviral therapy. Patients who have a favorable genotype can follow the current standard of care; for those with a less favorable genotype, doctors can recommend waiting for FDA approval of direct-acting antiviral drugs to improve their chances of response.

"I'm very impressed with the research Scripps is doing and glad to be able to contribute to it," says Loretta Roberts, a study participant. "The more information they find out about each viral type, the more it will help other patients in the future, including my son who contracted hepatitis C during my pregnancy."

Later this year, Scripps researchers plan to offer an additional test to accurately predict anemia in hepatitis C patients taking the pegylated interferon and ribavirin drug combination. Anemia is one of the most common side effects of the regimen; this new genetic test will enable doctors to individualize therapy to prevent this problematic side effect.



Paul Pockros, MD

"This is a huge step forward in the movement toward individualized medicine. As a physician, knowing which drug therapies will have benefit and which ones won't based on a patient's genotype is a significant breakthrough. Now, we are able to target each patient's treatment."

-Paul J. Pockros, MD, Clinical Director of Research
Scripps Translational Science Institute

As part of a clinical trial for hepatitis C patients, Loretta Roberts has received genetic testing to determine that for the best response, she needs a combination of antiviral therapies.