



# InTranslation



NEWS FROM SCRIPPS GENOMIC MEDICINE

ISSUE 1 OCTOBER 2007

## Finding the Healthy Elderly

Do Jerman (pronounced Doe) has been active all her life and often jokes about having been born on the third hole of a golf course. She swing dances, works out with a personal trainer and, of course, plays golf. Jerman is part of an exclusive club: the well elderly. Though 81 years old, Jerman has had no significant chronic medical conditions, which gives her longevity particular significance. Researchers at the Scripps Genomic Medicine program want to know more about what keeps her so healthy.

### In the Genes?

You might think Jerman simply has good genes. In other words, she lacks the genes that could make her susceptible to conditions like diabetes or heart disease. After all, both her parents lived into their 90s, despite drinking, smoking and eating poorly. But researchers have learned that the well elderly often have a similar profile of disease-producing genes but, for some reason, still don't get sick. If you are wondering why that is, so is Dr. Eric Topol, director of the Scripps Health Translational Science Institute and Scripps Genomics Medicine program. Dr. Topol has initiated the "Welllderly" Study to answer that question.

"Everybody has bad genes, that's what's fascinating. But why do these people who harbor bad genes do so well?" asks Dr. Topol. "We need to find the mechanisms that keep them healthy, and we know these are lurking in the genome. We know there are 'modifier genes' that cancel out the risk of disease genes, but little work has been done so far in human beings. We're going to find these genes that protect from disease—they are really nature's secrets."

### Where Do You Look?

Locating the healthy elderly to get DNA samples is an interesting challenge, since they don't spend much time in hospitals, nursing homes or other care facilities.

"The logistics are tricky, but we have to find them," says Dr. Topol. "They don't walk into the clinic every day because



Do Jerman was eager to volunteer for the Welllderly Study.

they're so healthy. We're going to have to go out to the community to find them."

The genomics team found Do Jerman, or rather Jerman found them, when Dr. Topol presented at Scripps Health Foundation's Heritage Circle Dinner. Jerman was so impressed by Dr. Topol and the goals of the genomics program that she quickly decided to volunteer.

"He said he was looking for 80-year-olds on no medication to put in his gene pool. And I thought, here I am," says Jerman. "I wanted to do whatever I could to help."

### The Magic Number

Dr. Topol's team needs around 1,000 healthy volunteers age 80 or older to donate their DNA to complete the study. The process is simple: participants give a small amount of blood—around two tablespoons. This one-time donation will be added to the program's gene bank and, ultimately, used to determine what is protecting the healthy elderly from their "bad" genes—perhaps helping to develop future medical treatments or even cures.

"We are very excited," says Dr. Topol. "This is a unique study that could have a powerful impact on health."

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*If you are interested in participating in the Welllderly Study, or know someone else who might be, please call **1-800-SCRIPPS (1-800-727-4777)**, weekdays, 8:30 a.m.–4:30 p.m.*



Dear Friends:

Unraveling the mysteries of the human genome

is, without question, the most exciting project I have ever taken on. Using an array of incredibly sophisticated gene-reading equipment, we will begin to answer fundamental questions about human biology. Why do some people stay healthy despite having genes that should make them susceptible to disease? Why do blockbuster medications work on some people and not on others? How does ancestry influence health?

With the help of world-class researchers like Drs. Nicholas Schork, Kelly Frazer and Sarah Murray, Scripps Genomic Medicine hopes to answer these questions and many more. In addition, we are developing partnerships with other research organizations, like the J. Craig Venter Institute, biotechs and pharmaceutical companies to hasten the pace of discovery and translate that knowledge into new treatments.

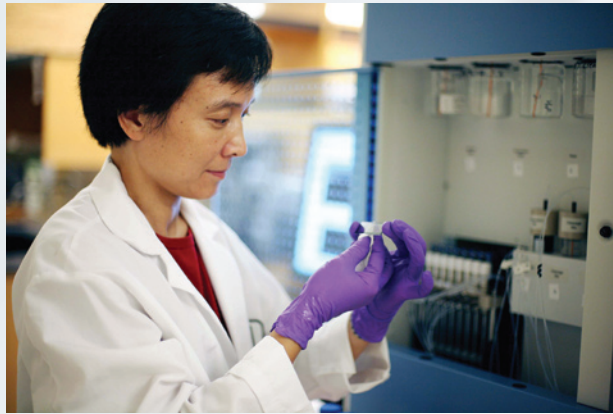
On this page is a brief description of our programs and what we hope to achieve in the coming years. We believe that, with our expertise and first-rate facilities, we will be important contributors to some of the coming era's most significant health care advances.

Eric J. Topol, M.D.

Chief Academic Officer,  
Scripps Health

Director, Scripps Genomic  
Medicine & Scripps Translational  
Science Institute

# New Programs for a New Era



Research Associate Wendy Wang at the Solexa genome analyzer.

## Scripps Genomic Medicine

Scripps Genomic Medicine is working to discover and define the genes that underlie susceptibility to disease.

There are more than 20,000 genes within 3.1 billion coding letters in the human genome and even more variations. With so many variables, how do you go about identifying which marker on what gene in

which chromosome impacts a specific disease? The key is narrowing down which genes and which markers are worth looking at and why.

In collaboration with The Scripps Research Institute, Scripps Genomic Medicine is creating a landmark biorepository for genetic research and study. Working hand-in-hand with volunteers from the more than half million individual patients we care for each year, Scripps is genotyping and storing genetic data on thousands of individuals of diverse ancestry. Once cataloged, the genetic information can be used to identify and better define those genes that play vital roles in human disease and medical conditions. These findings are taken directly into drug discovery programs, gene-based clinical trials and patient care.

## Scripps Translational Science Institute

The goal of the institute is to move discoveries from laboratory benches to patient bedsides in the safest, most efficient and most effective way possible.

A central part of the Genomic Medicine continuum, the institute analyzes genetic markers within the Scripps biorepository to identify promising research opportunities. Once identified, the institute advances clinical research and trials within the Scripps network and provides seed funding for physician researchers.

## Scripps Individualized Medicine and Clinical Trials

In collaboration with pharmaceutical, medical device, biotechnology and medical diagnostic companies, Scripps is reinventing the nature of clinical trials. Our individualized clinical trials capitalize on the latest in genetic technology.

Instead of testing 10,000 people with a scattered approach, genetic knowledge is improving our aim. Using individual genetic data, our highly advanced clinical trials specifically target small groups of 500 or less who share a particular gene variant.

Scripps Individualized Medicine and Clinical Trials is poised to bring the promise of genetic discovery full circle. Genetic data is used to identify potential research opportunities, which lead to highly targeted clinical trials and a shorter time between laboratory discoveries and their application in patient care. This elegant matching of an individual's genetic makeup with precise, gene-based medications and devices will lead to treatments that are generally safer and more effective than what is common practice today.

# Catching a Wave of Change

## Local Philanthropist Funds Genotyping System

John Gilchrist gives to Scripps in many ways. As a patient, he has trusted Scripps for his health care for more than 25 years and, as a philanthropist, he has invested in the organization by funding the purchase of leading-edge equipment to enhance care. When Gilchrist met Dr. Eric Topol and learned more about the Scripps Genomic Medicine program, he made up his mind to support this groundbreaking research.

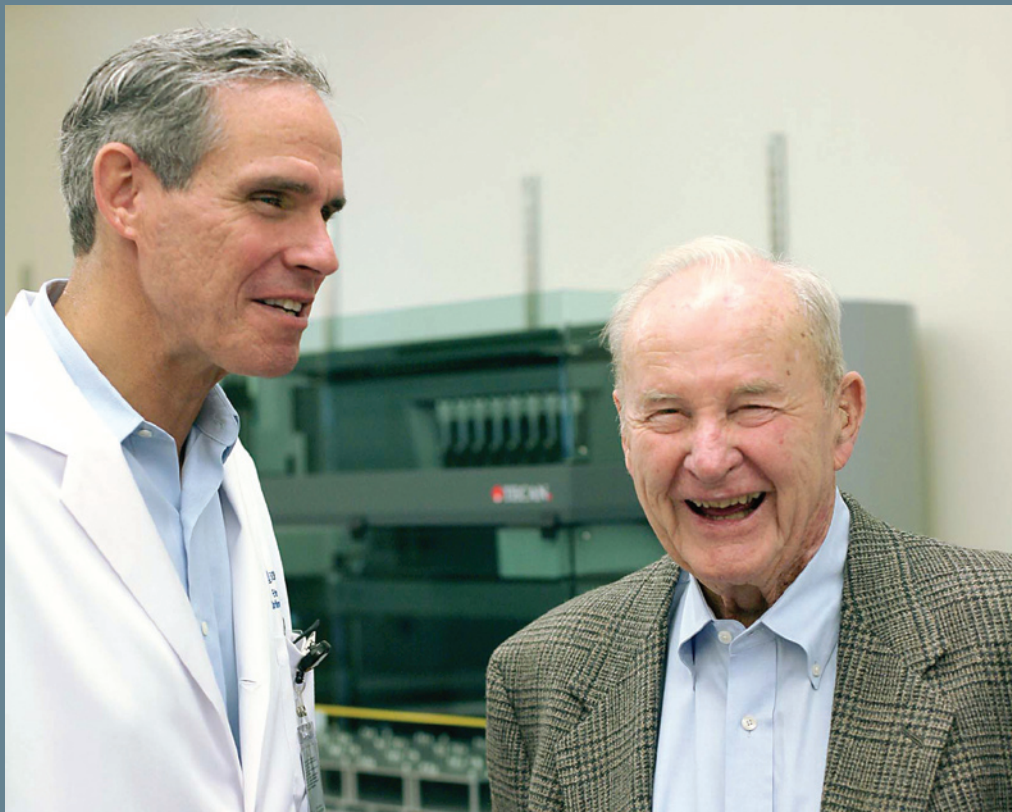
“Scripps is on the crest of a wave of change in medicine with the work that Dr. Topol is doing in the field of genomic medicine,” says Gilchrist. “The research is really miraculous, and I can’t think of a better way to help out.”

Gilchrist, a retired executive vice president for Coldwell Banker in the commercial real estate division, says genomics has him looking at medicine in a whole new way.

“Through the years, I’ve had several pacemakers implanted for my heart arrhythmia,” says Gilchrist. “Someday the genomic research going on will alleviate the need for treatments like this by discovering the underlying cause of the disease—and preventing it.”

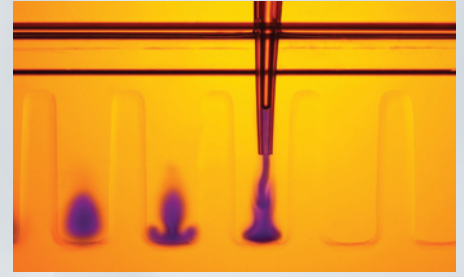
His recent gift will help fund the purchase of a \$1.2 million genotyping system that Dr. Topol and his team will use for large-scale analysis of genes, known as gene sequencing. By identifying mutations in genes that are responsible for certain diseases, the equipment will aid researchers in their work with the goal of ultimately preventing many diseases in the first place.

“It seems like every day there is a new discovery in the field of genomics,” adds Gilchrist. “Dr. Topol needs community support. My hope is that other people will join us today in creating a new era in medicine.”



John Gilchrist, pictured with Eric Topol, M.D., recently made a gift to help purchase the next generation genotyping system. The invaluable tool will aid Dr. Topol and his research team in their efforts to identify abnormalities in genes that can cause disease.

## Support the New Era of Medicine



Join us in creating a new world of medicine. Through your philanthropic support of the Scripps Genomic Medicine program, you will help us positively affect the lives of millions of people. You will accelerate understanding of the most fundamental mechanisms of life-threatening diseases and encourage the development of more effective treatment methods. And you will help assure that many serious health conditions will be a thing of the past.

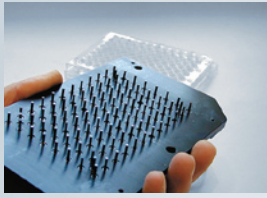
Scripps Health has provided significant funding to develop the genomic medicine program, but your help is vital to our efforts. We are a not-for-profit health care provider and receive no taxpayer subsidies. As private insurers continue to drastically curtail the portion they pay for health care, we have to look for additional ways to fund important programs and services. In addition, funding for medical research is at a standstill. Last year, the National Institutes of Health budget was cut by \$33 million.

Philanthropic investment is needed to help us transform the practice of medicine. Your financial support is an opportunity to be among a select group of people who can say they are part of a new era of medicine.

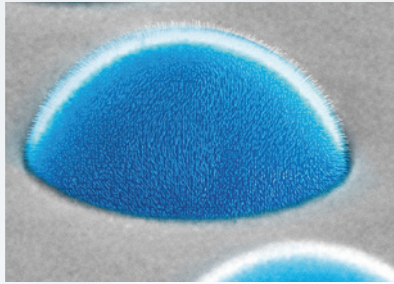
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*For more information on ways to give to the Scripps Genomic Medicine program, please contact Denise M. Scalzo, senior director of development, Scripps Genomic Medicine, at 858-554-3046.*

# Tools of the Trade



Scripps Genomic Medicine is working to answer some of the most profound questions about human genetics, but how do they do it? BeadChips, built by local biotech Illumina, are microarrays that use three micron (a micron is one millionth of a meter) silica beads to capture, examine and decipher specific parts of a genome. Each bead is covered with hundreds of thousands of copies of a specific oligonucleotide, a piece of DNA that captures complementary segments of DNA. Tests are then carried out on the captured segments of DNA to determine which DNA basepair occurs at a specific site in the genome. By analyzing which beads have captured specific DNA nucleotides in many samples, researchers can survey the majority of the genome and determine if there are any variations or other parts of the genome associated with disease.



# G-Nomics

**Gene**—The fundamental unit of heredity. A gene is an ordered sequence of molecules located in a particular position on a particular chromosome that encodes a protein or other molecule.

**Genetics**—The study of inheritance patterns for specific traits.

**Genome**—All the genetic material in the chromosomes of a particular organism; its size is generally given as its total number of base pairs.

**Genomics**—The study of genes and their function.

**Genotype**—The genetic constitution of an organism, as distinguished from its physical appearance.

**Genetic screening**—Testing a group of people to identify individuals at high risk of having or passing on a genetic disorder.

**Genetic testing**—Analyzing an individual's genetic material to determine predisposition to a particular health condition.

Courtesy of the Human Genome Management Information System,  
U.S. Department of Energy Human Genome Program



## SCRIPPS GENOMIC MEDICINE

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