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Kaiser Permanente, UCSF Scientists Complete NIH-Funded Genomics Project Involving 100,000 People

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Resulting Data Will Provide Novel Resource for Accelerating Research

OAKLAND, Calif. — One of the nation's largest and most diverse genomics projects has reached its first major milestone in just 15 months. Scientists have genotyped the DNA and analyzed the length of chromosome tips in more than 100,000 Kaiser Permanente members who agreed to be part of the research.

The project is a novel resource for the world of health science research because it will provide scientists with high-quality, genome-wide genetic data on a large and diverse population, according to lead investigators Cathy Schaefer, PhD, executive director of the Kaiser Permanente Research Program on Genes, Environment and Health (RPGEH); and Neil Risch, PhD, co-director of the RPGEH and director of the UCSF Institute for Human Genetics.

"By funding this project, the National Institutes of Health has significantly accelerated research into conditions such as cardiovascular disease, diabetes, cancers, mental health disorders, and age-related diseases such as Alzheimer's disease," Schaefer said.

The genetic information generated by the project will also include data about drug metabolism and drug response, which may help researchers to discover genetic factors that explain differences in the way people respond to medications. This would in turn help doctors provide patients with the best medicines for them individually, with less trial and error, based on their genetic background. It could also help researchers understand why some patients with cancer or heart disease, for example, develop certain symptoms and other patients do not, insights that may lead to new treatments and, in some cases, new ways to lessen the severity of or even prevent disease, Schaefer added.

The project has now completed its first phase, which was to extract and genotype DNA from a cohort of 100,000 participants with an average age of 65 and measure the length of participant telomeres — tiny units of DNA that bind the ends of chromosomes. Telomere length may reflect the degree of aging in a person's cells and may be a marker for age-related conditions.

Results of the genotyping and telomere length analysis will be linked to a broad spectrum of California environmental data and to current and historical health-related information from participant health surveys and the Kaiser Permanente electronic health record, the world's largest civilian electronic health record, the researchers said.

Risch said the pace of the project was only possible because of the alignment of a number of critical factors: Kaiser's Permanente's extensive health records and thousands of willing participants from the diverse Northern California region; new genotyping technology from Santa

Clara, Calif.-based Affymetrix, Inc., which produces scalable genomic analysis tools; and a history of collaboration between Kaiser Permanente and UCSF, two research institutions with complementary strengths for this project.

“The truth is that this project would have been impossible at any other time or place,” Risch said. “No single institution could have combined this level of genetic science with such deep health records on this diverse and large a number of patients. And without the funding, we could never have developed the technology to make this happen.”

The genotyping project was made possible by a two-year, $24.8 million grant awarded in September 2009 to the Kaiser Permanente RPGEH and UCSF by the National Institutes of Health. Funding for the grant came from three sources: the National Institute on Aging, the National Institute of Mental Health, and the Office of the Director.

”The completion of genotyping on this large and diverse population marks an unprecedented milestone in population-based genetics research,” said Richard J. Hodes, MD, director of the National Institute on Aging at the National Institutes of Health. “The genetic information, combined with the longitudinal clinical and health data that are already part of the Kaiser Permanente database, along with California environmental data, will create a unique and powerful resource to help answer research questions about aging, health and disease.”

To complete the genotyping project in two years, as required by the funding, Kaiser Permanente first had to build a new, high-throughput laboratory in Oakland, Calif., to extract DNA from more than 100,000 saliva samples in 15 months. The extracted DNA was transferred to UCSF’s Institute for Human Genetics, which worked with Affymetrix, Inc., a life science technology leader, to create custom Axiom® arrays for genotyping 675,000 to 900,000 markers, comprised of single nucleotide and insertion-deletion polymorphisms, across all 100,000 samples.

“This project reflects the incredible advances that have occurred in the past decade in the field of genomics and the speed and cost-effectiveness of genotyping technology,” said UCSF professor Pui-Yan Kwok, MD, PhD, who led the SNP genotyping work at UCSF. “Three years ago, we could never have undertaken a project of this size, much less completed it in only 15 months.”

Separate samples of DNA from the same 100,000 individuals were also analyzed in the laboratory of UCSF Professor Elizabeth Blackburn, PhD, who received the Nobel Prize in Physiology or Medicine in 2009 for her discovery of telomeres. The Kaiser Permanente/UCSF project will create the largest resource of telomere data available and will be the first such resource linked to health records of this number and diversity through Kaiser Permanente.

Over the next year data from the genotyping project will be processed and cataloged by RPGEH and UCSF scientists so that it can be made available to researchers in late 2012. For more information, visit www.rpgeh.kaiser.org

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