# Scripps Wellderly Genome Resource now available to researchers

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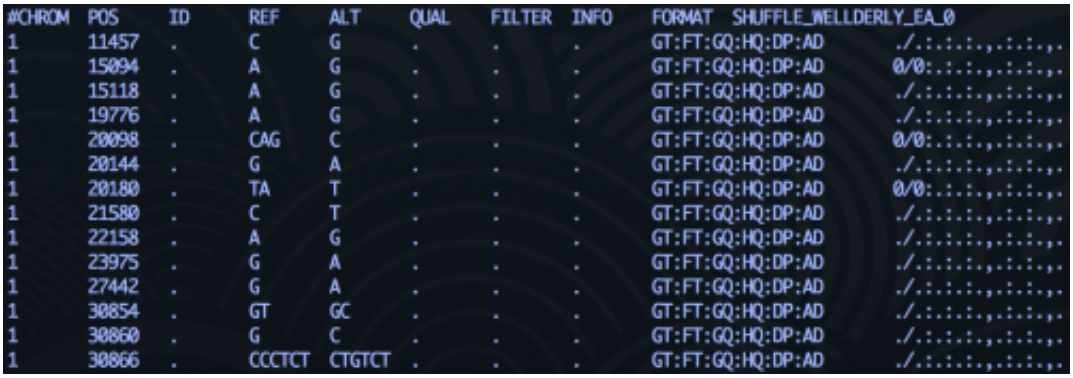

| #CHROM | POS | ID | REF | ALT | QUAL | FILTER | INFO | FORMAT | SHUFFLE_WELLDERLY_EA_0 |
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| 1 | 27442 | . | G | A | . | . | . | GT:FT:GQ:HQ:DP:AD | ./.:.:.:.,.:.:.,. |
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| 1 | 30866 | . | CCCTCT | CTGTCT | . | . | . | GT:FT:GQ:HQ:DP:AD | ./.:.:.:.,.:.:.,. |

This is a visual display of data from the Scripps Wellderly Genome Resource. Credit: Scripps Health

Scientists exploring the genetic causes of illnesses such as Alzheimer's disease, heart disease, cancer and diabetes now have a new tool – a reference DNA dataset built by researchers at Scripps Translational Science Institute (STSI).

STSI researchers announced the release of the Scripps Wellderly Genome Resource (SWGR) today at the Future of Genomic Medicine Conference, the institute's annual educational gathering of the nation's leading genetics experts.

The SWGR is comprised of the whole genome sequences of 454 participants in the Wellderly Study, an ongoing STSI research project

that is searching for the genetic secrets behind lifelong health by looking at the genes of healthy elderly people.

What makes this dataset stand out from other genomic references is the exceptional healthspan of the Wellderly participants, who have lived at least 80 years without developing any chronic diseases. Since other reference genomes include DNA data from younger participants, there is no way of knowing whether their disease status will change later in life.

Because the SWGR contains information from a healthy population, it can serve as a control group for a wide range of genomic studies of most late-onset adult diseases.

"We hope the Scripps Wellderly Genome Resource will play an important role in providing the research community extensive genomic characterization of a vital phenotype – healthspan – which has not been previously available," said Eric Topol, M.D., director of STSI and chief academic officer of Scripps Health. "We will continue to build on this with diploid, phased genomes in larger numbers of our Wellderly cohort."

Scientists around the world can now access the SWGR free through the San Diego Supercomputer and the financial support of a National Institutes of Health Clinical and Translational Science Award.

"The Scripps Wellderly Genome Resource offers the genomics community a best-case scenario for the presence of a DNA variant in a population," said Erick Scott, M.D., a clinical scholar at STSI and doctoral student in the labs of Ali Torkamani and Peter G. Schultz at The Scripps Research Institute in San Diego. Scott led the effort to analyze the Wellderly Study data.

Researchers who tap into the Scripps dataset can use another tool

developed by STSI called SG ADVISER. The free online search engine, located at genomics.scripps.edu/ADVISER/, delivers rich amounts of information about gene variants, including whether the searched-for variant appears in the Wellderly population and how frequently it shows up in that group.

The first published use of the SWGR came in January when a group of U.S. and U.K. researchers linked rare mutations of a certain gene to a twofold increase in the risk of developing late-onset Alzheimer's. The Scripps dataset served as a control for comparing members of 14 families in which the disease was common with a group lacking the illness.

The SWGR also has been used by STSI researchers who are studying the DNA of people with mysterious, or idiopathic, diseases that have defied more conventional diagnosis and treatment. The researchers are searching for rare genetic mutations that are related to the puzzling conditions.

"The SWGR will become more robust with time," said Ali Torkamani, director of genome informatics at STSI and an assistant professor of integrative, structural and computational biology at The Scripps Research Institute.

"As more DNA sequences from Wellderly Study participants are completed, their genomic data will be added to the Scripps Wellderly Genome Resource to strengthen the power of this innovative research tool," he said.

Provided by Scripps Health

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