

New collaboration combines genomic sequencing and stem cell technology to understand disease

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Today, the New York Stem Cell Foundation (NYSCF) Research Institute and PersonalGenomes.org announced a partnership to identify genetic and environmental contributions to trait and disease development. Cell lines generated by NYSCF will complement genomic data and medical histories contributed by participants in the Harvard Personal Genome Project (PGP), creating a unique and powerful resource to help researchers identify causes of disease.

"Mapping human genetics has laid the groundwork for personalized medicine to tailor treatments to patients on a level not previously achievable. And, now with [stem cells](#), we can take this a step further: we can test and refine drugs on a patient's actual diseased cells," said Susan L. Solomon, CEO of NYSCF.

"Our challenge, going forward, is to leverage the tremendous amount of information from the PGP as the world's only open access source of human genome, microbiome and disease data into a resource for testing causes and cures. We are using PGP stem cells for studying human mutations, gene editing therapies, and novel transplantation methods. That is why we see such value in the integration of these new stem cell technologies through this partnership with NYSCF," said George Church, PhD, Professor of Genetics Harvard Medical School and Founder of the PGP.

To achieve this goal, NYSCF scientists will generate stem [cell lines](#) from skin samples of participants in the Harvard PGP. These cell lines can then be studied and compared to data gathered by the PGP including whole genomes, medical histories, body microbiomes and hundreds of other traits from over 3,000 participants. This tool will help achieve the joint goal of "functionalizing" personal genetics for all individuals, meaning using personal genetic information to make informed medical and health decisions.

In 2001, the Human Genome Project (HGP) finished the first draft of the human genome, sequencing most of the 3 billion base pairs that compose human DNA. This took over ten years and cost nearly \$3 billion. Today, [human genome](#) sequencing is higher quality and can be completed in a day for a little over \$1000 enabling researchers an unprecedented look at the building blocks of both normal development and disease.

To additionally speed up the pace of discovery, the PGP developed the open consent framework which enables research studies to make genetic and medical data freely available online to the public and for researchers to analyze and begin to tie underlying genetic patterns or sequences to the development of traits and diseases. Similarly, NYSCF makes cell lines available to researchers in its efforts to find treatments and cures for disease.

Initially, 50 PGP participants will donate skin samples for generation of induced pluripotent stem (iPS) cell lines, which are genetically matched, self-renewing cells that can become any of the body's cell types. Clinicians at dermatology clinics will perform a punch biopsy, a minor procedure to obtain a skin sample, on consenting participants.

These skin samples are then sent to the NYSCF Research Institute, where scientists will derive iPS cells using the NYSCF Global Stem Cell

Array, a novel robotic technology that automates the generation of iPS cell lines. Unlike traditional, by-hand methods to procure stem cells, the Array creates standardized, quality controlled cells, enabling scientists to compare stem cells from different participants.

NYSCF scientists will take these iPS cells and derive different adult cell types of interest for research investigations. These cells, which reflect the participants' genetics, provide a powerful tool to study how genetic differences between people can affect disease development and trait expression. Additionally, NYSCF will make these [cells](#) available to the broader scientific community through the NYSCF repository.

"Overlaid with medical data, these stem cell models provide a more complete picture of each participant, effectively functionalizing stem cell technology," said Scott Noggle, PhD, Director of the NYSCF Laboratory and NYSCF – Charles Evans Senior Research Fellow for Alzheimer's Disease.

Provided by New York Stem Cell Foundation

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