Researchers create cell models of rare and undiagnosed diseases

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In what is anticipated to be a major step forward for rare disease research, The New York Stem Cell Foundation (NYSCF) Research Institute is partnering with the National Institutes of Health (NIH) Undiagnosed Disease Program (UDP). NYSCF scientists will generate stem cell lines from 100 patients in the UDP and collaborate with UDP researchers to better understand and potentially treat select rare diseases. While working collaboratively under this agreement, neither NIH nor NYSCF will receive funding. This partnership will leverage investments made using government (NIH) and private (NYSCF) dollars to ensure that research into rare diseases will move forward efficiently and effectively.

In 2008, the NIH developed the UDP to study patients with rare diseases, track their progression and symptoms, and obtain biological specimen samples to help pinpoint the root cause of their ailments. These patients, whose disorders have long-eluded diagnosis, provide skin samples that will be used to generate stem cell lines on the NYSCF Global Stem Cell Array™, a robotic technology that automates the derivation process. "The NYSCF Global Stem Cell Array™ provides us with the critical ability to produce accurate cell models of these rare diseases, serving as 'windows onto disease,'" said Susan L. Solomon, CEO of NYSCF.

"This collaborative effort has the potential to create new disease models that will reveal whether candidate gene variants are responsible for a patient's disease," said Dr. William Gahl, Director of the NIH UDP. "In

addition, the stem cells can illustrate new mechanisms with broad applicability to more common disorders." Gahl also noted that this venture may eventually be expanded to other centers within the Undiagnosed Diseases Network. The network is a group of 5 to 7 clinical sites, in addition to the UDP, that the NIH Common Fund will establish within the coming year.

NYSCF scientists will create induced pluripotent stem (iPS) cell lines by reprogramming UDP patient skin cells into an embryonic-like state. These iPS cells carry the same genetic information as the patient and can become any of the body's cell types. Scientists at NYSCF and the NIH will use these stem cells to model diseases in the petri dish and enable more comprehensive analyses of their underlying causes. Critical to research investigations, the iPS cells produced by the Array are completely standardized, enabling scientists to compare these cells to a patient's adult cells and relevant medical data using a platform that is reproducible and scalable.

Disease modeling, a novel application of stem cell technology, enables researchers to examine the molecular-level features of diseases in vitro, or in the petri dish. Many cell types are not readily accessible from patients and post-mortem examination inherently fails to show active disease processes necessary to better understand the ailment. IPS cells, when turned into adult cell types, play out the disease in a manner similar to that of cells in the patient. Unlike engineered animal models or cultured adult cells, iPS cells are patient-specific, readily comparable to the medical data of diseased patients from which they are derived.

NYSCF is using the Array technology to develop a collection of diverse stem cell lines that will be replicable and available to the scientific community for research use. This technology is particularly important for rare diseases, enabling the creation of a pool of stem cells to recreate the human disease model and conduct specified investigation into

unknown or little-studied ailments.

Consistent with the aims of the UDP, NYSCF and NIH scientists will collaborate on studies with these quality-controlled, standardized stem cells in a joint effort to advance medical knowledge about both common and rare diseases with the goal of providing answers to mysterious medical conditions and, ultimately, researching cures.

Provided by New York Stem Cell Foundation