

Stem cells for first time used to create abnormal heart cells for study of cardiomyopathy

June 9 2010

Researchers at Mount Sinai School of Medicine have for the first time differentiated human stem cells to become heart cells with cardiomyopathy, a condition in which the heart muscle cells are abnormal. The discovery will allow scientists to learn how those heart cells become diseased and from there, they can begin developing drug therapies to stop the disease from occurring or progressing. The study is published in the June 9th issue of Nature.

The Mount Sinai team used [skin cells](#) from two patients with a [genetic disorder](#) known by the acronym LEOPARD syndrome. Hypertrophic [cardiomyopathy](#), or thickening of the heart muscle, is experienced by 80 percent of patients with LEOPARD syndrome and is the most life-threatening aspect of the disorder. The Mount Sinai team took patient skin cells and reprogrammed them to become pluripotent [stem cells](#). Such cells can then develop into almost any type of cell in the human body. The researchers then created heart cells that had characteristics of hypertrophic cardiomyopathy.

"We knew there was potential in using [pluripotent stem cells](#) from people with genetic disorders to develop diseases in vitro, but our study is the first to successfully create abnormal heart cells," said the Principal Investigator of the study Ihor R. Lemischka, PhD, Professor, Gene and Cell Medicine, Developmental and Regenerative Biology, Mount Sinai School of Medicine. "Now that we have developed these cells, we can

study why they become enlarged and develop treatments to prevent them from overgrowing."

Scientists know that genetic disorders occur because of a mutation in a protein signaling pathway called the RAS pathway, but they have been unable to determine precisely how this results in disease-associated problems like hypertrophic cardiomyopathy. The authors of the Nature study concluded that induced pluripotent stem cell-derived [heart cells](#) provide the required characteristics to precisely determine the pathology behind these disorders, and a foundation for studying treatment interventions.

"This discovery has broad-reaching implications for genetic diseases like LEOPARD syndrome and Noonan's syndrome," continued Dr. Lemischka. "We look forward to further studying these cells as a potential therapeutic target."

Provided by The Mount Sinai Hospital

Citation: Stem cells for first time used to create abnormal heart cells for study of cardiomyopathy (2010, June 9) retrieved 9 April 2024 from <https://phys.org/news/2010-06-stem-cells-abnormal-heart-cardiomyopathy.html>

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