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[Stem Cell] Biologists Generate Man-made Pluripotent Stem Cells to Aid Study of Pompe Disease
[Stem Cell] Biologists Generate Man-made Pluripotent Stem Cells to Aid Study of Pompe Disease (Chinese Version)

Academia Sinica Newsletter (2011/11/28) Assistant Research Scientist at the Stem Cell Program of Institute of Cellular and Organismic Biology/Genomics Research Center, Dr. Hung-Chih KUO and his colleagues recently successfully generated the world's first Pompe disease-specific induced pluripotent stem cells. These stem cells constitute a promising model through which to test drugs and disease markers for Pompe disease.

Pompe disease (also known as glycogen storage disease type II) is a genetically inherited disorder caused by mutations in the gene encoding the enzyme acid alpha glucosidase (GAA). Without treatment, most patients with the infantile-onset form of Pompe disease die by the age of 18 months. Current understanding of the progression of Pompe disease during development is still limited, partly due to the difficulty in obtaining proper cell specimens from patients. To develop efficient therapies, researchers need to discover ways to obtain a more thorough understanding of the development of Pompe disease at the cellular level.

Pluripotent stem cells are unique among the different types of cells found in the body in that they are able to differentiate into a myriad of other types of cells, a characteristic known as "pluripotency". So called "induced pluripotent stem cells" (iPSCs) are man-made pluripotent stem cells that can be generated from any individual, including individuals with sporadic and inherited genetic diseases. These cells resemble human embryonic stem cells in many aspects. However, the generation of iPSCs from diseased cells (such as those found in the infantile form of Pompe disease) is challenging, as reprogramming efficiency may be compromised by defects caused by the diseased nature of the cells.

Dr. KUO and his colleagues generated iPSCs specific to Pompe disease and showed that these cells possess human embryonic stem cell characteristics and pluripotent developmental propensity. Furthermore, they demonstrated that these Pompe disease iPSCs are able to give rise to cardiomyocytes, (a type of specialized cell found in heart muscle) which exhibited the characteristics of Pompe disease. Drug rescue assessment revealed that the diseased cardiomyocytes could be rescued by various drug treatments including rhGAA, L-carnitine, and 3-MA. In addition, marker genes whose expression robustly correlated with the therapeutic effect of the drug treatment were identified.

The research was published in the advance access online edition of the journal Human Molecular Genetics on September 28. The full-text of the study entitled "Human Pompe disease-induced pluripotent stem cells for pathogenesis modeling, drug testing and disease marker identification" is available at the Human Molecular Genetics journal website at: http://hmg.oxfordjournals.org/content/early/2011/09/28/hmg.ddr424.full

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