

Scientists Induce Pluripotent Stem Cells From Pompe Disease Patients

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AsianScientist (Nov. 30, 2011) - A research team lead by Dr. Hung-Chih Kuo of the Academia Sinica in Taiwan has successfully induced the world's first pluripotent stem cells from the skin cells of Pompe disease patients.

In new research published in the journal *Human Molecular Genetics*, Dr. Kuo and his colleagues from the Institute of Cellular and Organismic Biology/Genomics Research Center generated iPSCs specific to Pompe disease and showed that these cells possess human embryonic stem cell characteristics and pluripotent developmental propensity.

Pompe disease, also known as glycogen storage disease type II, is caused by a mutation 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.

Although researchers have previously generated iPSCs from differentiated somatic cells, generating iPSCs from diseased cells such as those found in the infantile form of Pompe disease remains challenging, as reprogramming efficiency may be compromised by defects caused by the diseased nature of the cells.

Dr. Kuo's team showed that when the Pompe disease iPSCs differentiate to cardiomyocytes - specialized cells found in heart muscle - the cells exhibit the pathological characteristics of Pompe disease.

Next, the researchers tested drugs on the Pompe iPSCs and identified marker genes whose expression robustly correlate with the therapeutic effect of the drug treatment.

Having a cellular model for Pompe disease has many promising implications; it may someday enable early diagnosis, novel drug testing, and the identification of disease markers for the disease.

The article can be found at: [Huang HP et al. \(2011\) Human Pompe disease induced pluripotent stem cells for pathogenesis modeling, drug testing and disease marker identification.](#)

Source: [Academia Sinica](#).

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