Stem Cells Application in Modeling of Human Genetic Diseases

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Abstract

The use of animal models in modeling of human genetic disease has many advantages. In some cases, however, this method may not be applicable due to some limitations, such as differences in tissue composition, anatomy and physiology of humans and animals. Isogenic human disease models are a population of cells that are selected or engineered to model a specific genetic disease, in vitro. They are provided with a genetically matched 'normal cell' to provide an isogenic system. These cell lines are tailored by means of homologous gene targeting using targeting vectors. Two common cell sources for this purpose are hESCs and iPSCs. The use of iPSCs technology began in 2007. In this technique, a blood sample or a skin biopsy is taken from the patient, and then by using the reprogramming inducing factors, iPSCs are generated. Since most disease phenotypes are only observed in differentiated cells, but not in stem cells, iPSCs must be differentiated into disease-relevant cell line. On the other hand, in order to create an isogenic system, normal cell line is to be generated by homologous gene targeting. Making use of stem cells, cellular models of many genetic diseases have been developed so far. Lesch-Nyhan syndrome (using hESC) and Huntington disease (using iPSC) are just two examples in which cellular models has been successfully applied to display a disease phenotype. Using cellular models has enabled researchers to better understand the genetic diseases mechanisms and to evaluate the effects of novel therapeutic agents. Moreover, this method could be used to predict which particular patient groups would better respond to a particular drug treatments and enhance drug development, leading to personalized therapies.

Key words: Cellular model, Genetic disease, Stem cells, Reprogramming.

Poster Presentation

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