TECHNOLOGY

Induced Pluripotent Stem Cells Derived from Patients Diagnosed with Gaucher Disease

OVERVIEW

Gaucher disease (GD) is a genetic disorder in which glucocerebroside accumulates certain organs because the body fails to produce sufficient quantities of lysosomal glucocerebrosidase (GCase). The mechanisms by which GCase deficiency causes GD is not understood due to the limited availability of relevant tissues from affected patients. To overcome these limitations, human induced pluripotent stem cells (hiPSC) from GD patients harboring the GCase mutations have been generated. The GD derived hiPSC cells have been shown to differentiate into macrophages and neuronal cells that express the phenotypic and pathological varients of the disease. The research tool can be used to understand molecular mechanisms and developing therapeutic approaches for GD.

Skin biopsies were collected from Gaucher disease patients to generate hiPSC of types 1, 2 and 3 of GD. HiPSC differentiated into macrophages and neuronal cell types. The hiPSC derived macrophages exhibited the functional defects of types 1, type 2, and type 3 GD.

APPLICATIONS

There are three types of GD. GD Type 1 is the most common of the disease in western countries, making up 95% of the patients. GD Type 1 is treatable. GD Type 2 is rare, involves severe neurological abnormalities, and is usually fatal within the first two years of life. GD Type 3 is the most common form of the disease worldwide. Type 3 has symptoms of both Type 1 and Type 2. With treatment, Type 3 patients can live into their 50s.

The most effective therapy for Type 1 and the non-neurological symptoms of Type 3 is enzyme replacement therapy (ERT). ERT involves bi-monthly intravenous infusions. ERT cannot be used to treat the neuropathy in types 2 and 3 GD patients. Substrate reduction therapy is a newer treatment that works by reducing the amount of glucocerebroside that the body makes.

Though GD is a rare disease, the prevalence is one in every 40,000 live births in the general population and 1 in 450 live births in individuals of Ashkenazi Jew (Eastern European) descent. The average cost for ERT treatment is approximately $250,000/year with an annual revenue of about $1.25 billion.

ADVANTAGES

Can be used for Gaucher disease modeling and drug screening

STAGE OF DEVELOPMENT

A sensitive cell-based assay is currently under development for the evaluation of therapeutic efficacy of new drugs that may have dual use for GD and Parkinson’s Disease.

(Updated 10/2017)

Additional Information

LICENSE STATUS

Available for licensing; Available for sponsored research

INVESTIGATOR(S)

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ATTACHMENTS

- Download RF-2012-043 summary sheet-final.pdf

EXTERNAL RESOURCES

- Induced pluripotent stem cell model recapitulates pathologic hallmarks of Gaucher disease.
- Gaucher iPSC-derived macrophages produce elevated levels of inflammatory mediators and serve as a new platform for therapeutic
- Gaucher Disease-Induced Pluripotent Stem Cells Display Decreased Erythroid Potential and Aberrant Myelopoiesis

RF-2012-043