Induced Pluripotent Stem Cells Derived from Patients Diagnosed with Gaucher Disease

Summary

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.

Key Investigator
Ricardo Feldman

Field
Tangible Research Tool

Technology
Gaucher Disease model iPSC

Advantages
Can be used for Gaucher disease modeling and drug screening

Status
Available for licensing
Available for sponsored research

UMB Docket Reference
RF-2012-043

External Reference
Proc Natl Acad Sci U S A. 2012 Oct 30; 109(44): 18054–18059.

Stem Cells. Author manuscript; available in PMC 2015 Sep 1.

Stem Cells Transl Med. 2015 Aug; 4(8): 878–886.

Market

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.

Technology

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.

Technology Status

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.

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