The FSH Society of the United States and the FSHD Canada Foundation of Calgary, Alberta, Canada announce first joint effort to fund FSHD research using human induced pluripotent stem cell (hiPSC) technology that could lead to new insight into the FSHD disease process while providing critically important tools for developing new therapies.

Dr. Gabsang Lee
(Johns Hopkins University, Baltimore, Maryland, USA)

Derivation of human induced pluripotent stem cells from FSH patient fibroblasts
(2013-2014)
$49,705 over 1 year

[Provided by Dr. Gabsang Lee:] “The genetic and biological events that result in Facioscapulohumeral muscular dystrophy (FSHD) pathogenesis are complex and the link between the genetic aberration and manifestation of symptoms is still elusive. We hypothesize that there might be cellular and genetic alterations in the early stage of myogenesis in FSHD patients. The establishment of human induced pluripotent stem cells (hiPSCs) ushered in a new era in biomedicine and can be useful for modeling pathogenesis of human genetic diseases, autologous cell therapy after gene correction, and personalized drug screening. Our lab has been studying human genetic disorders by using induced pluripotent stem cells (hiPSCs), a new type of stem cell generated without destruction of any embryonic tissues or embryos. In addition, we already built a novel methodology in a highly innovative manner to directly derive and prospectively isolate skeletal muscle from the hiPSCs. Here we propose to establish hiPSC lines from FSHD patients’ somatic cells. Our proposed study will enable us to isolate FSHD-specific skeletal muscle cells for better understanding of FSHD pathogenesis in the human system as well as for advancing potential autologous cellular therapy to correct genetic defects in FSHD in the near future.”

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