



Induced Pluripotent Stem Cell Lines for Fragile X Syndrome Research

WARF: P160138US01

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The Wisconsin Alumni Research Foundation (WARF) is seeking commercial partners interested in developing human induced pluripotent stem cell (iPSC) lines to enable in vitro modeling of FXS.

Overview

Fragile X syndrome (FXS) is a genetic disorder characterized by mild to moderate intellectual disability and a characteristic phenotype. It is the most common form of inherited intellectual disability and is closely linked to autism. FXS is caused by an expansion of the CGC triplet repeat within the 5'-untranslated region of the FMR1 gene on the X chromosome, which causes a deficiency in the resulting FMR protein that is needed for normal brain development. There is no cure for FXS, and research is still needed to understand the full molecular and cellular consequences of this mutation of the FMR1 gene.

The Invention

UW-Madison researchers have created human induced pluripotent stem cell (iPSC) lines from fibroblasts obtained from individuals with FXS to enable *in vitro* modeling of FXS. iPSC lines from three FXS patients were created along with one isogenic disomic control. Additionally, mosaicism of the CGC repeat length in one patient's fibroblasts allowed for the generation of isogenic cell lines with differing CGC repeat lengths from that patient. The researchers have confirmed that the FMR1 mutation is preserved while reprogramming the fibroblasts to iPSCs, and the FXS forebrain neurons differentiated from the iPSCs display defective neurite initiation and extension.

Applications

- Model system for researching the mechanisms underlying FXS and the molecular and cellular consequences of FXS
- Potential usage in drug screening to treat FXS and autism

Key Benefits

- Well-characterized resource for examining potential neuronal deficits caused by FXS and the function of FMRP in human neurons
- Patient-derived stem cells to study FXS under the human genetic background
- Few FXS-specific stem cell lines exist

Additional Information

For More Information About the Inventors

- [Anita Bhattacharyya Consigny](#)
- [Su-Chun Zhang](#)

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