



Induced Pluripotent Stem Cell Lines For Fragile X Research

WARF: P160138US02

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The Wisconsin Alumni Research Foundation (WARF) is seeking commercial partners interested in developing cell lines for Fragile X syndrome research and drug screening.

Overview

Fragile X syndrome (FXS) is the most common form of inherited intellectual disability and is closely linked with autism. The genetic basis of FXS is an expansion of CGG repeats in the 5'-untranslated region of the FMR1 gene on the X chromosome leading to the loss of expression of the fragile X mental retardation protein (FMRP). The cause of FXS has been known for over 20 years, yet the full molecular and cellular consequences of this mutation remain unclear. Although mouse and fly models have provided significant understanding of this disorder and its effects on the central nervous system, insight from human studies is limited.

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 the human disease. Three young boys with FXS who came from a well-characterized cohort representative of the range of affectedness typical for the syndrome were recruited to aid in linking cellular and behavioral phenotypes. The FMR1 mutation is preserved during the reprogramming of patient fibroblasts to iPSCs. Mosaicism of the CGG repeat length in one of the patient's fibroblasts allowed for the generation of isogenic lines with differing CGG repeat lengths from the same patient. FXS forebrain neurons were differentiated from these iPSCs and display defective neurite initiation and extension. These cells provide a well-characterized resource to examine potential neuronal deficits caused by FXS as well as the function of FMRP in human neurons.

Applications

- Patient-derived stem cell lines
- Model system to study disease processes of FXS under the human genetic background
- Drug screening for FXS

Key Benefits

- Well-characterized fragile X patient specific induced pluripotent stem cell lines
- Biomaterial readily available for licensing

Stage of Development

The induced pluripotent stem cell lines including FX11-01-7, FX13-01-2, FX 08-01-01, and FX11-01-9U, were submitted to WiCell for banking and distribution December, 2014. The materials have already been expanded and tested at WiCell, and are ready for distribution.

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For More Information About the Inventors

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Tech Fields

- [Research Tools : Cell lines](#)

For current licensing status, please contact Andy DeTienne at adetienne@warf.org or 608-960-9857

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