



## Induced Pluripotent Stem Cell Lines for Down Syndrome Research

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**The Wisconsin Alumni Research Foundation (WARF) is seeking commercial partners interested in developing Ts21-induced pluripotent stem cells (iPSCs) as a model system for studying Down syndrome.**

### Overview

Down syndrome (Trisomy 21, Ts21) is a condition caused by having an extra copy of the 21st chromosome due to an error in cell division. People born with Down syndrome have a characteristic phenotype and many potential associated disabilities. Although Down syndrome is the most common genetic cause of intellectual disability, the precise molecular mechanisms underlying the associated cognitive impairment are not clearly understood. The study of these underlying mechanisms has been limited by the lack of a model system that contains full trisomy of chromosome 21 in a human genome that enables normal gene regulation.

### The Invention

UW-Madison researchers have created Ts21-induced pluripotent stem cells (iPSCs) from two sets of Ts21 human fibroblasts. Four iPSC lines have been created, three with Ts21 and one isogenic control that is disomic for chromosome 21. It has been confirmed that the Ts21 iPSCs and neurons display developmental defects that are consistent with the cognitive defects seen in individuals with Down syndrome. Differentiation of the Ts21 iPSCs yielded similar numbers of neurons expressing markers characteristic of dorsal forebrain neurons that were functionally similar to controls, and expression profiling revealed changes that are consistent with the presence of 50 percent more genetic material of chromosome 21.

### Applications

- Model system for studying Down syndrome in a human genetic background
- Potential platform for drug discovery to treat complications of Down syndrome
- Potential discovery of underlying causes of Down syndrome

### Key Benefits

- Disease cell lines with an isogenic control
- Few Down syndrome-specific stem cell lines exist

### Additional Information

#### For More Information About the Inventors

- [Anita Bhattacharyya Consigny](#)
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