

## GENOMES AND CEREBRAL PALSY

### Identifying drug-based treatments for genetic cerebral palsy

Cerebral palsy is an umbrella term that refers to a group of disorders affecting a person's ability to move. It is caused by damage to the developing brain either during pregnancy or shortly after birth.

Until recently only 2% of cerebral palsy cases were thought to be genetic.

Research now shows that up to 45% of cases may have a genetic cause. Thanks to remarkable advances in genetic sequencing of the human genome, researchers can now find genetic variations that are the likely cause of CP in some people.

More research is required to understand the role of 'CP genes' as a cause of CP, and to develop genomically guided treatments for these type of CP.

Recent groundbreaking findings have led us to establish a collaborative, international CP genomics network to harness several emerging genomic technologies, and undertake coordinated studies to enhance our understanding of CP genomics.

Over 5 years this project will explore ways to restore normal cell function in children who have genetic based cerebral palsy.

#### PROJECT OUTLINE

##### Discovering the genomes

How can a single change in a child's genetic code profoundly alter brain development and lead to CP?

Such findings are crucial to understanding normal brain development, as well as how CP develops when this process goes awry. Using state-of-the-art genome sequencing technology and powerful computing resources, researchers at Phoenix Children's Hospital will investigate genetic code from hundreds of people with CP in order to find a single genetic variant that leads to cerebral palsy.

##### Understanding the impact on brain cell structure and function

How do genetic mutations affect cell function?

Using induced pluripotent stem (iPS) cells (reprogrammed blood or skin cells) researchers will then investigate specifically how genetic mutations impair brain cell structure and function, which in turn can lead to cerebral palsy.

This will then enable the investigation of new drug-based therapies that have the potential to restore normal cell function in people with cerebral palsy.

##### Preclinical Testing

Are potential drug-based treatments both safe and effective?

Before new drug treatments can be safely administered to patients they require thorough testing in animal models to ensure effectiveness and safety. Successful preclinical testing will see this project move to human clinical trials.

#### RESEARCHER

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**FUNDING REQUIRED**

**\$3 million over 5 years**