10,000 people in world-first cerebral palsy study

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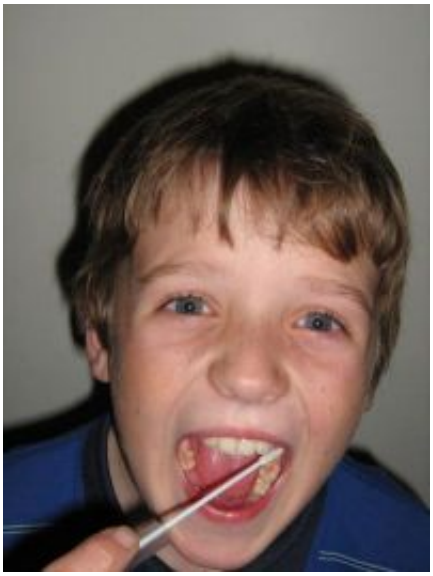

Nine-year-old Elliott demonstrates how to do a cheek swab. Credit: Photo by Michael O'Callaghan

Researchers from the University of Adelaide, Australia, have launched the largest study of its kind in the world in a bid to better understand the possible genetic causes of cerebral palsy.

The study – requiring cheek swabs of mothers and their children – aims to gather genetic samples from 10,000 people right across Australia.

One of the world's most serious complications during pregnancy and

birth, cerebral palsy is a disability that affects one in every 500 children worldwide, and the consequences are life long.

Over the next two years the researchers will test 5000 participants from families affected by cerebral palsy, while the other 5000 without an affected child will consist of a control group.

"Our study will investigate a key issue behind cerebral palsy: whether genetic factors make women more vulnerable to environmental risks that affect the brain of their unborn child. These risks – such as prematurity and infections – combined with genetic susceptibility mean that babies could be at double jeopardy of cerebral palsy," says research leader Professor Alastair MacLennan, Head of Obstetrics & Gynaecology at the University of Adelaide . PhD student Michael O'Callaghan is the national coordinator of the trial.

"Recent studies by our group suggest that cerebral palsy may be associated with genetic and other mutations that may increase blood clotting within the brain," says Professor MacLennan, who is also head of the South Australian Cerebral Palsy Research Group, the world's leading research group into the causes of cerebral palsy.

"An association between cerebral palsy and different types of herpes virus infection – such as cold sores and chicken pox – has also been discovered in South Australian studies.

"The next step is to see if this is true in a much larger population, comparing the genetics of both mother and child," he says.

People with cerebral palsy lack control of their movement and posture as a result of brain injury in the neuro-motor region. The symptoms vary greatly in severity, ranging from poor muscle co-ordination to quadriplegia.

Cerebral palsy is usually present from birth. The injury to the brain does not get worse over time.

"It was once thought that cerebral palsy was caused by low oxygen levels during birth. However, this is rarely the case," Professor MacLennan says.

"Obstetric care and caesarean deliveries have increased six-fold over the last 50 years, but the incidence of cerebral palsy cases has remained the same. Most of the cases are associated with problems during pregnancy and possible genetic susceptibility. Currently there is no cure or way to prevent cerebral palsy," he says.

"If our research confirms that there are genetic mutations that can lead to cerebral palsy, specific disease preventions may be available for individuals.

"In the future, gene therapy may allow doctors to alter the aberrant genes in a mother or fetus, or specific drugs could be used to counter the effect of genetic mutations and ultimately prevent a child from developing cerebral palsy before birth.

"Knowledge of a patient's genetic makeup and tailored administration of anti-inflammatory drugs before and during pregnancy may be possible. Immunisation against viral infections also may be a future option when this preventative therapy is available," he says.

Source: University of Adelaide