MedicalXpress

Research shows how genetic mutation causes epilepsy in infants

October 3 2007

New research from the Howard Florey Institute in Melbourne has shown why mutation in a single gene can cause epilepsy in infants.

Infants are more susceptible to seizures because their brains are developing at a rapid rate, making their brain cells 'excitable'. Their neurons are growing and making new connections with other nerve cells, which can disrupt normal brain activity and results in epilepsy.

Infants have protective mechanisms in their brains to control this excitability, but now the Florey scientists have uncovered that a single gene mutation prevents a specific ion channel from functioning correctly, thus causing excitability which results in epilepsy.

Dr Steven Petrou and his team knew the genetic mutation existed, but did not know its impact on brain function or that it may control excitability in infants' brains.

"This discovery is helping us to understand how and why this form of epilepsy, known as benign familial neonatal-infantile seizures, appears in these infants," Dr Petrou said.

"Interestingly, we also found that the ion channel which carries the mutation is itself naturally protective as it limits excitability in the infant brain by waiting to fully 'switch on' only in the adult brain.

"The mutation accelerates this normally delayed development change,

leaving the susceptible infant brain with an overly excitable channel and epilepsy.

“If this switch to a more excitable state occurs too early during brain development, it is possible that epilepsy and other neurological disorders develop.

“This highlights the complexity of the human brain and how disorders can develop if one single mechanism goes awry,” he said.

This research was recently published in Molecular and Cellular Neuroscience. According to Dr Petrou, potential new avenues of research may emerge from this study.

Epilepsy affects 3% of the Australian population and better treatments are urgently needed as many existing therapies have serious side-effects for patients. In 30% of cases, the current drug options available provide limited seizure relief.

There are more than 200 types of epilepsy, which can either be inherited or result from a range of causes including, head trauma, vascular disease or brain tumours.

Source: Research Australia