

## **New treatment possibilities for fatal genetic disease**

### **National Huntington's Disease Awareness Day is Friday 30 March**

Researchers at Melbourne's Howard Florey Institute have opened up new treatment possibilities for Huntington's disease by proving a scientific theory incorrect.

It was previously thought that dopamine neurons, called D2 neurons, were responsible for the devastating symptoms seen in Huntington's disease but the Florey researchers have proven that loss of D1 neurons causes many of the fatal diseases disabling symptoms.

In Huntington's disease, the first evidence of damage in the brain occurs in the part of the brain called the striatum; D1 and D2 neurons constitute 90% of neurons in the striatum.

Research leader, A/Prof John Drago, said now that the importance of D1 neurons in HD had been established, they could work towards therapies that focused on both D1 and D2 neurons.

"Currently there is no effective treatment for Huntington's disease and patients suffer from debilitating movement, memory, and psychiatric problems," he said.

A/Prof Drago's discovery was made after he genetically engineered a mouse that displayed Huntington's disease features by damaging only D1 neurons.

While a mouse model that carries the human Huntington's disease gene already exists, A/Prof Drago's mouse model is the first in the world to accurately mimic the death of the D1 neurons in the striatum.

"Despite the widespread death of D1 neurons, the mouse was healthy, apart from having HD symptoms," A/Prof Drago said.

"This indicates that there is potential for a tremendous amount of natural repair occurring in the Huntington's diseased brain."

"Now the challenge is to thoroughly understand how this natural repair occurs so we can develop a therapy that encourages and enhances repair in human patients."

"Using the brain's own adult stem cells to naturally repair and prevent further damage is one treatment possibility that we eventually hope to explore," he said.

In addition to his research career, A/Prof Drago is a neurologist at the St Vincent's Hospital Movement Disorder Clinic, where he treats patients with HD.

"It was the lack of effective treatments for patients that inspired me to undertake this research, so it is extremely satisfying to solve another piece of the Huntington's disease puzzle and work towards a cure for this progressive genetic disease," he said.

A/Prof Drago's research was published in the 26 February 2007 edition of the highly prestigious *Proceedings of the National Academy of Sciences* journal.

He was assisted by Dr Ilse Gantois, a postdoctoral researcher from Belgium, and the Florey's Neuroimaging group led by A/Prof Gary Egan, who undertook MRI scans of the mouse model to show its shrinking striatum as D1 neurons were dying and the response of the brain by making glial cells.

The Florey is taking two different approaches to its Huntington's disease research with this investigation and also Dr Anthony Hannan's environmental enrichment research, which has shown

that physical and mental stimulation, can delay the onset of the disease and slow the progression of symptoms.

By tackling HD from two different angles, the Florey researchers hope to accelerate their discoveries into clinical outcomes to benefit HD patients.

### **Huntington's disease**

Huntington's disease (HD) is a genetic disorder, passed on from parent to child, which results in a variety of devastating symptoms that worsen as the disease progresses. Each child of a parent with the disease has a 50-50 chance of inheriting the HD gene and therefore developing the disease.

### **Statistics and Facts**

- HD affects approximately 7 people per 100,000 of the population in Australia
- HD tends to appear between the ages of 30 and 50
- The Huntington's gene is dominant, meaning that a person who inherits it will inevitably develop the disease
- Evidence suggests that the prevalence of HD is highest in populations of Western European background
- One of HD's earliest names was chorea, the Greek word for dance

The Howard Florey Institute is Australia's leading brain research centre. Its scientists undertake clinical and applied research that can be developed into treatments to combat brain disorders, and new medical practices. Their discoveries will improve the lives of those directly, and indirectly, affected by brain and mind disorders in Australia, and around the world. The Florey's research areas cover a variety of brain and mind disorders including Parkinson's disease, stroke, motor neuron disease, addiction, epilepsy, multiple sclerosis, autism and dementia.

### **ENDS**

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