

What is Huntington's disease?

Huntington's disease is caused by the deterioration of various brain regions, including the cerebral cortex and basal ganglia, and causes uncontrollable body movements, and stiffening of limbs. It can also cause emotional and cognitive impairments, including dementia, loss of memory, judgement and awareness.

Although there is an increasing number of people diagnosed with Huntington's disease without any family history, it is a genetic condition that a child inherits from their parent. Only one parent needs to carry the gene mutation that causes the condition, and the children of affected parents have a 50/50 chance of inheriting the disease gene.

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If the gene mutation is inherited, they will develop Huntington's disease.

Huntington's disease usually manifests when people are in their 40s or 50s. The earlier the symptoms appear, the faster the disease seems to progress. Huntington's disease is progressive, meaning it worsens over time. The expected lifespan for people with the condition once symptoms appear is 10–25 years. There is currently no disease-modifying treatment or cure for Huntington's disease.

In Australia, more than 1800 people live with Huntington's Disease, and over 9000 are at risk.



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The Huntington's disease gene includes a 'genetic stutter' that causes a 'tandem repeat disorder', where a sequence of three DNA nucleotides—cytosine, adenine and guanine, or CAG—are repeated too many times. When this DNA repeat is translated into protein it makes an abnormally long tract of the amino acid glutamine, repeated within the huntingtin protein. This polyglutamine tract is toxic to specific types of neurons and also other cells in the brain and body.

Huntington's disease is one of over 30 tandem repeat disorders which include other diseases as diverse as spinocerebellar ataxias, Kennedy's disease, fragile X syndrome, and a subset of motor neuron disease and frontotemporal dementia.

The expanded polyglutamine tract in the huntingtin protein can disrupt the expression of many other genes and impair the transport of proteins and other molecules around cells. It can also affect the function of synapses, which are crucial in brain circuits mediating various aspects of cognition, emotion and movement.

Huntington's disease research currently involves a wide range of disease models, as well as clinical studies. Much of the research focuses on how specific molecules and cells are disrupted, and how they give rise to the movement disorder, psychiatric symptoms and cognitive deficits, which culminate in dementia.

Some regions of the world have unusually high rates of Huntington's disease, such as Lake Maricabo in Venezuela, and Tasmania in Australia.



The field is rapidly moving to establish sophisticated models of disease mechanisms, develop new treatments, and to establish clinical trials.

As Huntington's disease is a well-defined brain disorder involving a specific gene mutation, it is ideal for neurotechnology innovation. Neurotechnology applications could be used to prevent or ameliorate the movement disorder, psychiatric symptoms and cognitive problems.

The Australian Brain Initiative will nurture the basic brain research required to better understand and effectively treat disorders like Huntington's disease.

The Initiative will also progress collaboration between research and industry to advance neurotechnology devices that have the potential to transform not only the capabilities of neuroscience research but also the lives of those living with conditions and disorders of the brain.

Huntington's disease causes swelling of the ventricles and atrophy of cerebral nerve tissue.



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