

Motor Neuron Disease

What is Motor Neuron Disease?

Motor Neuron Disease (MND) is the name given to a group of related brain disorders that affect the motor neurons in the brain and spinal cord, leading to weakness and muscle wasting. It results from the steady but inexorable loss of nerve cells that control muscle movement and function (motor neurons). Unfortunately, this loss may not become evident until many nerve cells have died; thus the patient may undergo rapid deterioration shortly after diagnosis, which ultimately results in death due to loss of the nerve cells that control breathing.

The cause of the majority (>95%) of MND cases is unknown. However, in a small percentage of cases the condition is genetic: the patient inherits an altered form of a gene, which ultimately leads to the disease. Significantly, there is currently no therapy to reverse the effects of the disease or to prevent its progression.

Research at QBI

In 2005, geneticist Dr Robyn Wallace was awarded the Ross Maclean Fellowship for further study into MND.

Dr Wallace is pursuing several avenues of MND research in her laboratory at QBI which include:

Gene expression

In collaboration with Tony Hannan (University of Melbourne) the Wallace laboratory is investigating how environmental enrichment can delay disease onset by analysing gene expression levels in a mouse model of MND.

Gene expression levels will also be examined in blood samples from MND patients to identify molecular markers associated with MND.

Mouse models of MND

A small proportion of MND patients carry mutations in the superoxide dismutase (SOD1) gene. Transgenic mice carrying the mutant SOD1 gene undergo progressive motor neuron loss. In collaboration with QBI's Dr Elizabeth Coulson, Dr Wallace's team is investigating ways of preventing cell death associated with MND in SOD1 mice.

ENU mutagenesis

Through the Australian Phenomics Facility, QBI has access to hundreds of mice carrying thousands of random-point mutations. Dr Wallace's plan is to screen these mice for loss of motor function to identify genes relevant to MND. Some mice have already been identified that warrant additional characterisation.

Improving diagnosis

MND can be difficult to diagnose, which is important for the subsequent management and care of the patient. Dr Wallace will soon be recruiting patients for a new research project which will determine whether biomarkers exist in the blood that are specific to MND. She will also track changes in these biomarkers in MND patients over time, to see if biomarkers can be used to predict the course of the disease.

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Fast facts

- 1,400 people in Australia are living with MND.
- Each day at least one Australian dies of MND and a new person is diagnosed.
- Average onset age is 59 years; however, the age of onset can range from 20 to 70 years.
- Although MND was first described nearly 150 years ago, there is still no known cause or cure.

Source: Motor Neuron Disease Association of Australia

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