FIGHT MND.

# Project

## Disease heterogeneity

### Developing a high-throughput system to identify MND risk genes

For most people living with MND, their diagnosis comes as a surprise. There is no family disease history, and the cause is unknown. Research, however, is uncovering that genetic causes of MND are much larger than previously recognised, and just this year, five new regions of the genome were found to contribute to MND. Investigators in this project will study these regions in more detail to identify the specific MND risk genes. Successful outcomes will identify new lead genes responsible for MND and new targets to treat MND.

*"A treatment that stops the disease in its tracks remains critical for those with sporadic MND."*

# Project Lead

## Dr Fleur Garton

### The University of Queensland, QLD

Dr Fleur Garton says although she has no specific favourite research project, she is inspired by the fact that ultimately *"research can prevent or cure a disease like MND"*.

In talking about research successes, she cites the incredible datasets that the international MND research community is now collating (including in Australia) and notes that continuing this momentum will be critical to producing a positive outcome for those living with MND.

## Risk genes

Dr Garton's lab is currently working to identify risk genes for MND. 'Risk genes' can alter the likelihood of an MND diagnosis and *"knowledge about how their regulation or expression can contribute to MND could help design a treatment,"* she says.

Her project is specifically focused on sporadic MND, which is the more than 80% of MND cases that do not have a family history or single causal gene variant identified.

To help identify these risk genes, Dr Garton and her team are *"looking at biomarkers (in the blood) and genetic variation (risk genes) associated with MND"*, a task that is assisted by human patient samples garnered from the Sporadic ALS Australia - Systems Genomics Consortium (SALSA-SGC) platform (also funded by FightMND).

She is hopeful that these research avenues may help diagnose, track, and ultimately treat MND and is excited that this project has real potential.

*"We are only at the cusp of risk gene discovery in ALS/MND, with many more to be investigated in the next few years. This project will help us streamline the process for these expected discoveries while also revealing new treatment avenues now,"* she says.

Dr Garton recognises that *"a treatment that stops the disease in its tracks remains critical for those with sporadic MND."*

*"Thanks to this funding from FightMND, we hope to reveal treatment avenue/s that can do this."*

**FightMND has invested $250,000 in this research.**

## About Dr Fleur Garton

Dr Fleur Garton works at the Institute for Molecular Bioscience at The University of Queensland. Her research experience spans both molecular genetics (testing gene function with knock-out/overexpression models) and complex trait genomics (the individual differences between people caused by genetic factors).