FIGHT MND.


Dr Fleur Garton

Researcher.
DR FLEUR GARTON

IMPACT Project.
A NOVEL BIOMARKER FOR ALS

Where do you work?

I work at the University of Queensland's Institute for Molecular Biosciences.

What is your research experience and background?

I began my research career in a neuromuscular research group based at Westmead Children's Hospital in Sydney under the guidance of Professor Kathryn North. My PhD investigated the genetic contribution of a variant involved in muscle function and following this, I moved with Kathy to Murdoch Children's Research Institute in Melbourne. I have been at the University of Queensland with Professor Naomi Wray since 2016 where the research has focused on understanding the genetic contribution to Motor Neurone Disease.

What led you to pursue your research into MND?

I have a passion for improving the lives of those affected by neuromuscular disease. MND is a devastating disease with a severe prognosis. Without a clear cause for many of those affected – I was keen to make a difference.

Can you describe the research your laboratory is currently focusing on?

We are focused on understanding the genetic contribution of MND. This includes tracking disease and we have begun an investigative study to look at cell-free DNA (short fragments of DNA floating in the bloodstream) to see if it can provide insight into mechanisms involved in MND and an easy way to diagnose or track the condition.

How did you identify that cell-free DNA has the potential to detect MND?

Cell-free DNA is a by-product of cells turning over. It is widely used in prenatal screening to analyse the baby's DNA using the mother's blood and is also being used in cancer – where tumour DNA can be assessed for causative mutations. Given MND is a rapid disease with many cells affected, we thought investigating the cell-free DNA profile was a worthwhile project to initiate and complement our other genetic and phenotypic investigations.

What excites you about cell-free DNA?

A simple blood test that can provide a snapshot of what is going on in the body would be a game changer for MND – cell-free DNA might help us in this domain.

What difference will this grant make to your work?

This grant will provide us with the means to collect and analyse cell-free DNA profiles from those with and without MND. Any findings can then be followed up in a larger cohort to see if they can contribute to the development of a robust biomarker test to diagnose and track MND. We are incredibly thankful for the FightMND organisation and its supporters for making this possible.

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Motor neurone disease (MND) currently affects ~2000 Australians, many of whom have spent over one year searching for a diagnosis. MND typically involves a rapid decline of function and there is no validated biomarker to speed up diagnosis, track or evaluate treatment effectiveness. The lack of a robust, objective biomarker is a critically missing element needed to evaluate MND clinical trials.

Cell-free DNA (cfDNA) may provide a rapid, efficient and sensitive solution to this problem. cfDNA is simply the product of cellular turnover, in which DNA originating from a cell's nucleus is detected in the circulating blood. cfDNA is increased in the blood of ALS patients (the most common type of MND) and can be profiled to recognise the originating tissue/cell. We propose that an MND/ALS specific cfDNA signature may be detectable during the disease course, ultimately leading to a blood test that could be requested at first presentation to a local clinic, or used to track disease progression.

OBJECTIVES:

To genetically profile blood-test-derived cfDNA samples from individuals presenting with MND symptoms and age-matched controls. Results from testing will be paired with a rich set of clinical data to ensure development of a robust and 'fit for purpose' test for the diagnosis, prognosis, prediction or tracking of MND.

OUTCOMES:

1. Development of a rapid, efficient, sensitive and economical way to speed up diagnosis and track the progression of MND/ALS with blood sample-derived genetic biomarkers. An early detection diagnostic screening test for MND would open the window to earlier therapeutic interventions, well before the disease has progressed.
2. cfDNA profiling will build a deeper understanding into the biology of ALS and provide unique insights into therapeutic avenues that have not yet been considered.
3. Advancement of a test that provides an accurate evaluation of MND clinical trials and the effectiveness of treatment.