FIGHT MND.

Project

Disease biomarkers

EC-FUS a novel biomarker for MND examined using a unique antibody.

Biomarkers are molecules that detect or confirm the presence of a specific disease. Currently, MND-specific biomarkers are not available for routine clinical use, which is delaying MND diagnosis for patients by up to 12 months. This project examines if a new pathological protein linked to MND, called EC-FUS, can be detected by patient blood tests to diagnose and measure the progression of MND.

Project Lead

Prof Julie Atkin

Macquarie University, NSW

Prof Julie Atkin says that her favourite personal research finding was the *"discovery of the mutations in C9orf72 in MND"* and this is because *"this gene is the most frequent cause of both MND and the related condition frontotemporal dementia"*.

She is also proud of her team who collectively were the first *"to describe the normal cellular function of the C9orf72 protein, which is significant in understanding MND"*.

Why it's hard to diagnose MND

Prof Atkin explains that MND is hard to diagnose as it can mimic other neurological diseases. There isn't a specific test or method that can confirm a diagnosis of MND. Instead, diagnosis typically involves a clinical examination and series of diagnostic tests that rule out other diseases that mimic MND.

"Diagnosis can take up to a year, which is far from ideal considering the short average survival time of MND patients following diagnosis," she says.

"This prolonged diagnostic delay also means patients cannot enrol early into clinical trials, thus preventing them from obtaining potentially disease-modifying treatments. Therefore, effective and accurate biomarkers are urgently needed to expedite the diagnosis of MND."

EC-FUS as a molecule for detecting MND

Prof Atkin's team discovered that EC-FUS was present in easily obtained biological fluids (serum, cerebrospinal fluids), as well as in MND patient spinal cords, *"where the levels of it are significantly different to individuals without neurological diseases"*.

"We recognised that it might have potential as a new biomarker to help diagnose and detect the progression of MND," she says.

FightMND has invested $249,972 in this research.

About Prof Julie Atkin

Prof Julie Atkin works at the Macquarie University Centre for Motor Neuron Disease Research, a recognised centre of expertise within the University. She is a cell biologist/biochemist and has worked predominately on MND for the last 18 years. Her research involves identifying the main cellular processes that trigger neurodegeneration in motor neuron cells and from this, designing new therapeutic approaches for MND.