

# Macquarie University Centre for Motor Neuron Disease Research



# Foreword

The Centre for Motor Neuron Disease Research exemplifies Macquarie University's commitment to doing quality research with real-world impact. The Centre's research in motor neuron disease (MND) and related neurodegenerative diseases is outstanding.



**Professor Patrick McNeil**

Deputy Vice-Chancellor  
(Medicine and Health)

Executive Dean, Faculty of Medicine  
and Health Sciences

Macquarie University is now widely known for MND research and clinical care, thanks to the hard work and dedication of this team of researchers, clinician-scientists and research students.

The quality of the Centre's research is illustrated by the recent award to two of its group leaders, Professor Ian Blair and Professor Gilles Guillemin, of a National Health and Medical Research Council (NHMRC) Investigator Grant, each worth more than \$2.2 million. These ultra-competitive grants are part of the NHMRC's new grant scheme for funding research leaders to pursue their ambitious discovery agendas.

The Centre is a flagship of MQ Health, the University's academic health science centre. MQ Health's vision is to integrate research and teaching with clinical services to deliver the highest quality healthcare to its patients.

The Centre for MND Research typifies MQ Health's mission by integrating the intensely personal care delivered to MND



**Professor Sakkie Pretorius**

Deputy Vice-Chancellor (Research)

patients by Professor Dominic Rowe and his team at MQ Health Neurology with leading research. In particular, the Centre has established a research pipeline that draws on patients' biological samples to further understand the causes of this horrendous disease and develop potential treatments.

What we find particularly exciting is that this intersection of clinical care and research is now delivering clinical applications. Professor Rowe is undertaking two clinical trials of potential MND therapies, testing existing drugs that are considered to have potential in MND now that we understand more of its underlying cellular and molecular causes. Moreover, potential therapies that have been developed within the Centre are undergoing pre-clinical evaluation.

It is an exciting time in MND research; we are finally beginning to see a glimmer of light at the end of this dark tunnel. We therefore commend the achievements outlined in this report and especially the Centre's vision for the next stages of its mission to beat MND.

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# Introduction

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In 2013, five separate and outstanding research teams took a leap of faith to relocate to Macquarie University with the collective desire to build a research powerhouse in motor neuron disease.

Central to this goal was integrating multiple scientific disciplines in a coordinated research 'pipeline' that engages patients in research. To this end, we established the Centre for MND Research, the Multidisciplinary MND Service and Clinic, and Australia's largest MND patient biobank. Working together, the Centre, the Clinic and the Biobank facilitate our breakthrough discoveries to better understand MND, accelerate new drug discovery and evaluate therapies in clinical trials.

Our strategy has sustained growth in MND research and catalysed new major national and international collaborations. We continue to attract and develop exceptionally talented, dedicated researchers who work closely with

the Clinic and are driven by the desperate need to beat MND.

Integral to the operation of the Centre is the care of people who have MND. We provide integrated care to more than 200 people with this disease, from initial diagnosis to multidisciplinary management of a complex and changing disease, to care in the community and end-of-life care.

There are no two people who have MND in the same fashion and, in much the same way, MND affects families of people living with MND very differently. The whole focus of our research effort is to change the survival and quality of life of people with MND. We use a patient-centric model of care to achieve

the best outcome for people with MND.

Until we can stop the progression, MND will continue to cause the death of more than 800 Australians each year.

As you will see in this report, our outcomes represent a team effort. From a standing start in 2013, we are now one of the largest MND research centres in the world, with multiple strands of funding from government and not-for-profit sources.

Thank you to all of those involved in bringing this report together. We look forward to sharing with you the progress in the battle to slow and stop this devastating disease. And, thank you to all of our generous donors who enable us to redouble our efforts to beat MND.



**Professor Julie Atkin**  
Centre Co-Director and  
Group Leader



**Professor Ian Blair**  
Centre Co-Director and  
Group Leader



**Professor Dominic Rowe AM**  
Centre Deputy Director

# Our vision

is a world without MND.

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# Our mission

is to provide world-class care for people living with MND and to develop new effective treatments for this devastating disease.

# Achievements 2018/2019



One of the world's strongest multidisciplinary hubs researching MND.

Produced **> 130**

original research publications in international peer-reviewed journals.



Set up Australia's first indigent fund for MND patients unable to access care.

**420**

Working with more than 420 global research partners.



Genetic testing results in babies born through IVF without a faulty MND gene.

Received **\$3.9 m**

in philanthropic funding.



Australia's largest longitudinal MND biobank.

Received **> \$5.8 m**

of grants and fellowships.

Home to more than **90**

researchers working on MND.



Launched and hosts the annual Macquarie Neurodegeneration Meeting.



Global studies to identify genes associated with MND.

Provided comprehensive care for **10%**

of Australians living with MND.



Conducted two clinical trials: CuATSM and the Lighthouse. The first site in the world to trial this therapy in MND.



Developing the next generation of leaders in MND research.

**4**

Applied for 4 patents based on research outcomes from the Centre.



Postgraduate student training program in MND research.

# Top minds join MND fight

In 2013, researchers sharing a dream to beat motor neuron disease (MND) came together from five institutes across Australia. With complementary expertise, they built a research centre at Macquarie University.

The Centre for MND Research combined a laboratory-based research pipeline with the Multidisciplinary MND Service and Clinic led by Professor Dominic Rowe. An expert in neurodegenerative disease and the inaugural Professor of Neurology at Macquarie University, Rowe started the clinic when Macquarie University Hospital opened in 2010. It was one of the first in Australia.

The new integrated centre set up Australia's largest longitudinal Neurodegenerative Disease Biobank for MND, one of the largest in the world. Patients and family members provide samples, including blood, skin, urine and hair, during regular clinic visits, all of which are stored and made available to researchers according to strict protocols. This enables the Centre to seek to understand

the disease's progression over time, as well as investigate causes and therapies.

Today, the Biobank is managed by Dr Sarah Furlong, while Professor Ian Blair, Professor Gilles Guillemain, Dr Angela Laird, Professor Julie Atkin and Professor Roger Chung lead five research teams that collaborate on the many projects the Centre runs. They also work closely with other institutions nationally and internationally, feeding research results back into the Multidisciplinary MND Service Clinic.

Since it was created, the Centre has produced more than 300 peer-reviewed journal articles and globally participated in the identification of 10 genes known to cause or confer risk to developing MND.

Highlights include publications in *Nature*

*Communications*, *Neuron* and *The New England Journal of Medicine*. Grant success has continued, including multiple NHMRC grants and fellowships, providing more than \$5.8m in funding during financial years 2017-18 and 2018-19.

The Centre has made breakthrough discoveries to understand MND and has multiple new potential drugs in development. The Centre has also hosted two clinical trials directed by Rowe. These include the CuATSM trial and the Lighthouse Trial, which is testing a novel antiretroviral therapy for MND and is the first site in the world to do so. The Centre also launched and hosted the annual Macquarie Neurodegeneration Meeting, a national conference attended by more than 150 delegates in 2018 and 2019.



Left to Right: Professor Gilles Guillemain, Professor Roger Chung, Dr Angela Laird, Professor Dominic Rowe, Professor Julie Atkin, Professor Ian Blair

# Our research teams

*'Success would look like us being able to slow and stop this disease that's robbing families of fathers, mothers, wives and husbands, sons and daughters ... If we can stop some deaths, that's success.'*

- Professor Dominic Rowe





NEURODEGENERATIVE  
DISEASE BIOBANK

# Frozen vaults fuel future research

The Neurodegenerative Disease Biobank is the largest facility in Australia dedicated to collecting and storing biological samples and clinical data for use in MND research.

Established in 2013, the Biobank is the cornerstone of the research program at the Centre for MND research. The facility holds more than 37,000 biological samples such as blood, urine, hair or skin.

Patients attending the Multidisciplinary MND Service and Clinic are invited to participate by donating their biological samples to the Biobank. Their family members and supporters are also invited to participate, to allow comparison between those with MND and those without. These samples are an invaluable resource for researchers to increase understanding of MND, and to identify treatments and preventative measures.

“We have an unusually high participation rate compared to other biobanks,” says Dr Sarah Furlong, who manages the facility. “Participants are keen to play a role in finding the cause of this devastating disease. The reputation of the research centre is a major factor in this.”

An advantage of the Biobank being located in the same building as the Clinic is that samples can be quickly processed and stored.

In addition to samples, the Biobank collects extensive demographic, lifestyle and clinical data, which is uploaded to a dedicated content management system. This increases the power of the research immensely.

“This is critical so that we can track biological changes with clinical changes,” says Furlong. “There is a lot of variability in MND patients. For example, some are initially affected in their speech, others by weakness in one of their limbs. Some first develop symptoms in their twenties, others in their eighties.”

So far, researchers have used more than 4,000 biobank samples, integrated with data, in 18 projects across the fields of genetics, proteomics, cellular stress, biomarkers and environmental studies. These projects include collaborators across Macquarie University, nationally and overseas.

The Biobank is part of an Australian consortium called the Sporadic ALS Australia Systems Genomics Consortium—SALSA-SGC. This consortium comprises nine MND centres across Australia working together to build an integrated infrastructure. Their focus is to ensure the centres follow global best practice standards in their data collection and storage.

### PROJECT MinE

The Biobank is also a member of a global collaboration, Project MinE, which aims to analyse the DNA of more than 15,000 MND patients and 7,500 control subjects from 20 countries. The aim of Project MinE is to identify what causes sporadic MND.

Samples are also sent worldwide for analysis for numerous genetics, proteomics, biomarkers and environmental studies.



NEURODEGENERATIVE  
DISEASE  
BIOBANK TEAM

Manager  
**Sarah Furlong\***

Technician  
**Kristine Deang\***

System Developer  
**Vimala Jacobs\***

Coordinator  
**Susan D'Silva\***

Biobank Technician  
**Elisa Cachia**

\* Pictured

### PROCESSING SAMPLES

Technicians adhere to strict protocols to ensure the best possible quality of samples and data – the facility was one of the first biobanks to obtain NSW Health Biobank certification, in March 2019.

A robotic instrument extracts DNA from blood rapidly and consistently. Other high-tech instruments measure sample quality.

Ultra-low temperature freezers maintain sample integrity by freezing the samples at as low as  $-196^{\circ}\text{C}$ .

Samples and data are coded to protect the privacy of the participants.



The collections are processed into eight different sample types: plasma, serum, deoxyribonucleic acid (DNA), ribonucleic acid (RNA), peripheral blood mononuclear cells (PBMCs), urine, hair and fibroblasts.



GENETICS AND GENOMICS TEAM

# Gene puzzles pieced together

Inherited gene mutations cause about 10 per cent of MND cases. The remaining cases are sporadic and the causes unknown. Worldwide, scientists have identified more than 20 genes that cause the disease. Further investigation may lead to therapies to slow MND progression.

As leader of the Centre's Genetics and Genomics team, part of a global consortium that has identified numerous inherited genes that cause familial MND, Professor Ian Blair is keen to understand the genetic factors that account for the variability in MND. These include the age when symptoms first appear and how fast the disease progresses.

"If we can identify which genetic or genomic factors are responsible, we can work to design therapies to delay progression of the disease and maximise the length and quality of life for MND patients," he says.

His team is also working to identify the gene variations that put people at risk of developing sporadic MND, whether it is combined with lifestyle choices or exposure to environmental factors. He is hoping this work may open up other avenues to develop therapies.

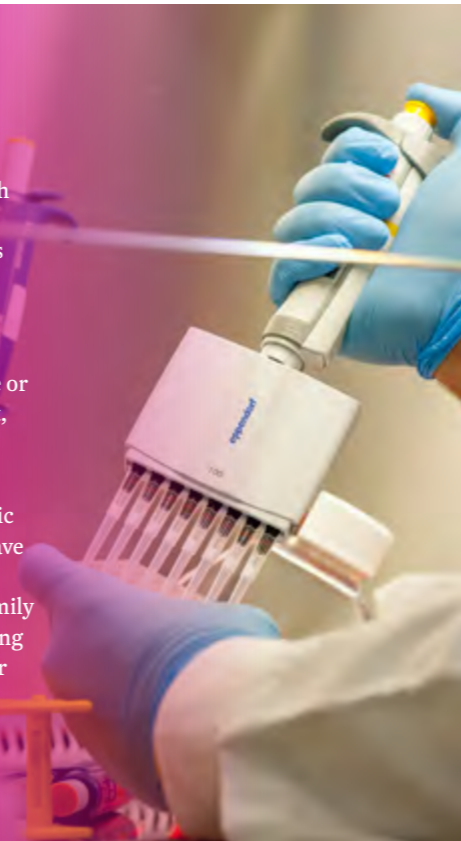
*"There are many ways to kill a motor neuron," says Professor Ian Blair.*

## GENETIC COUNSELLING

The genetic information Professor Blair's team identifies is channeled back into the Multidisciplinary MND Service and Clinic to inform the genetic counsellors who work with MND patients and their families. Finding a genetic cause for MND can have an enormous impact on individuals, he says.

"Families with MND may have up to 50 per cent of any one generation developing the disease; perhaps a brother, sister, aunt, uncle or parent died from the disease, and before that, a grandparent. So MND is very much in the forefront of their lives."

Those patients can undergo genetic diagnostic tests. Around 60 per cent of MND families have a known mutation. Early diagnosis informs clinical care and may extend life. "If these family members want to have children, genetic testing and IVF technology offers the opportunity for some to see these faulty genes stop at their generation," Blair says.



GENETICS AND GENOMICS TEAM

## GENOME SEQUENCING

Professor Blair's team is using DNA samples to sequence the entire genome, the three billion letters in the genetic code of MND patients, in Australia and also as part of an international consortium that seeks to identify the genetic and environmental factors that increase the risk of developing sporadic MND.

His team includes coding experts who use high-powered computers to decipher the genomes. This allows them to compare the genomes of MND patients and contrast them with those people without MND.

They can then spot unique changes in the code that cause MND or that make people susceptible to developing the disease.

The genetic discoveries inform the Centre's ongoing research, which seeks to understand how a motor neuron dies and to develop new therapies. When the team identifies a new genetic mutation, they can use it as a new tool to mimic the disease in the laboratory, put it into nerve cells or transfer into zebrafish to understand disease biology and test therapies that one day could translate to success in clinical trials.

Group Leader  
**Professor Ian Blair\***

NHMRC Early Career Research Fellow, Team Leader  
**Dr Kelly Williams\***

Postdoctoral Research Fellows, Team Leader  
**Dr Shu Yang\***

Postdoctoral Research Fellow  
**Dr Jennifer Fifita**  
**Dr Lyndal Henden\***  
**Dr Alison Hogan\***  
**Dr Emily McCann\***

Research Assistant  
**Natalie Grima\***

PhD Students  
**Sharlynn Wu**  
**Sandrine Chan\***

Masters Research Student  
**Winonah Riddell\***

\* Pictured





# Hunt for biomarkers and environmental triggers

Searching for biological markers to determine the rate of progression and severity of MND, and identifying possible environmental triggers, are the two main areas of focus for Professor Gilles Guillemin's team.

Professor Gilles Guillemin oversaw the study that identified the world's first biological marker, or biomarker, for Multiple Sclerosis in 2017. His research on neuroinflammation, neurotoxicity and the amino acid tryptophan is renowned internationally. Neuroinflammation is inflammation of the nervous system, including the brain and spinal cord. Neurotoxicity is when the nervous system is exposed to natural or manufactured toxic substances that alter its normal activity and/or kill nerves or brain cells.



## BIOMARKERS

Guillemin's team also analyses blood collected by the Centre's Neurodegenerative Disease Biobank, the largest facility of its type in Australasia. For more than two years, his team has received blood every six months from MND patients and their relatives (as controls) who attend the Multidisciplinary MND Clinic. This blood is then stored in the Biobank.

The team's aim is to identify new blood biomarkers able to predict how fast the disease is progressing—and whether patients respond to treatments or not. Dr Vanessa Tan is assessing more than 90 different molecules in the blood of these patients and comparing them to controls and analysing how they change over time.

## ENVIRONMENTAL FACTORS

The team is also investigating possible environmental triggers that may lead some people to develop MND.

Using data collected by MND Australia and the Multidisciplinary MND Clinic, Professor Dominic Rowe has identified 'hotspots' in NSW, where the incidence of the disease is up to five times higher than the average. One such hotspot is the town of Griffith in the Riverina of central New South Wales. The team is studying urine samples from about 30 people with MND living in the region and is actively seeking new participants for the study.

"Because Griffith is an agricultural area, we are checking for more than 50 different pesticides, for 10 metals such as iron, copper, mercury and arsenic—and for cyanotoxins—to find out why these people are developing MND more than the rest of the population," Guillemin says.



## MICROORGANISMS

Guillemin and Dr Benjamin Heng, in collaboration with the University of Sydney, have initiated a new research project looking at the possible roles of microorganisms (viruses, bacteria and fungi) as another potential environmental cause of sporadic MND.

Neurotoxicity is when the nervous system is exposed to natural or manufactured toxic substances that alter its normal activity and/or kill brain cells.



NEUROINFLAMMATION  
AND NEUROTOXICITY TEAM

## CYANOTOXINS AND MND

Cyanotoxins are created by cyanobacteria, commonly called blue-green algae, which bloom at warm temperatures in stagnant or slow-running water that contains high levels of nutrients, such as runoff from farmland.

In collaboration with other research teams based in the US, France, South Africa and Australia, the team is assessing whether beta-Methylamino-L-alanine (BMAA) and other cyanotoxins may trigger MND.

"The number of people dying from MND has doubled over the past two decades and this has interestingly paralleled the number of algal blooms in NSW in this period," Professor Guillemin says.

The team is using satellite imagery to try to match algal blooms with the incidence of MND.

Dr Vanessa Tan was part of the international team which identified that BMAA can propagate from neuron to neuron and other brain cells, and then become dysfunctional and progressively die. This could play a role in the development of MND for people exposed to frequent algal blooms and is the hypothesis of this current project.

Tan is also developing a new fluorescent BMAA so that she can more easily study the mechanism of BMAA propagation and neurotoxicity under a microscope in real time.

Group Leader  
**Professor Gilles Guillemin\***

Research Fellow  
**Dr David Lovejoy\***

Postdoctoral Research Fellows  
**Dr Benjamin Heng\***

**Dr Vanessa Tan\***

Research Officer  
**Dr Sharron Chow\***

Visiting Fellow  
**Dr Michelle Lima Garcez\***

Visiting Scholar  
**Dr Abid Bhat\***

PhD Students  
**Amanda Pires\***  
**Ali Ali\***

\* Pictured







NEURODEGENERATION  
TREATMENT TEAM

# Pathways towards drug treatments

Researchers within the Neurodegeneration Treatment Team are testing potential drug treatments on zebrafish. These fish are also helping the team better understand the degeneration of the nervous system that occurs in people with MND.

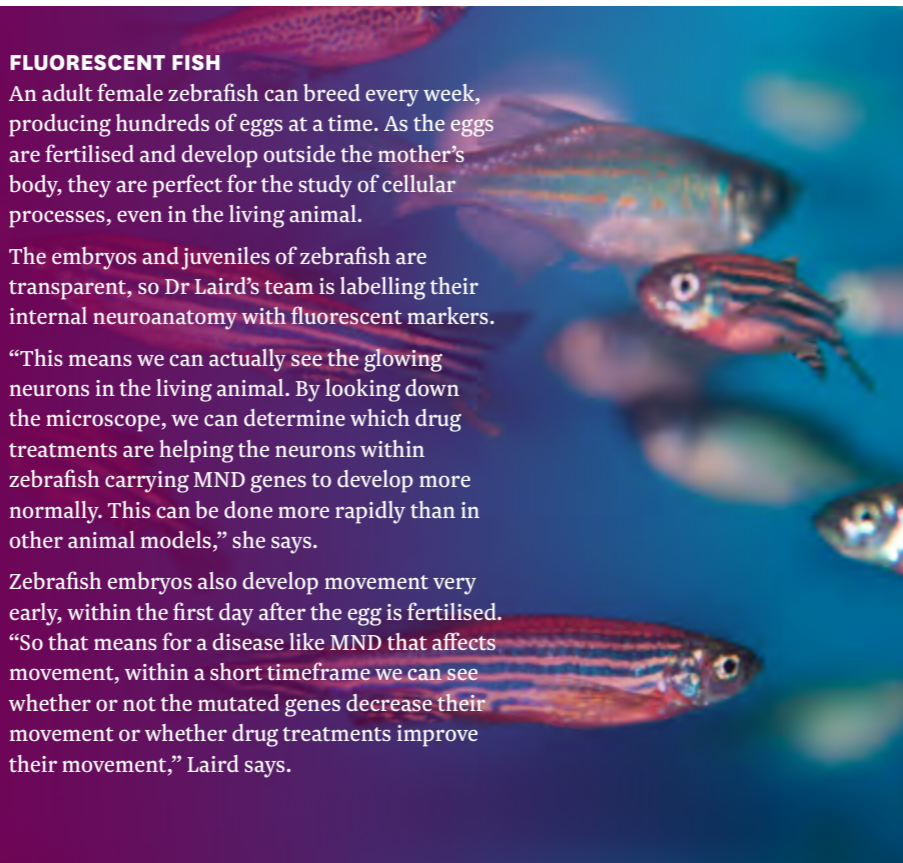
Zebrafish (*Danio rerio*) are an ideal tool to study drug treatments that may prevent or slow the development of MND. A vertebrate, they share 70 percent of their genetic code with humans and their embryos develop in days.

These tiny tropical fish can also absorb drugs added into the water they swim in, which makes it easy to dose fish that have MND with different drugs. "You can see quite quickly if it improves their movement," says group leader, Dr Angela Laird.

Prior to joining the Centre for MND Research in 2016, Laird used zebrafish to study neurodegenerative diseases with a leading MND team at KU Leuven, Belgium. She then established her own team at the University of Sydney's ANZAC Research Institute and led a project that produced and characterised the world's first zebrafish model of spinocerebellar ataxia type-3 (also known as Machado Joseph Disease, MJD).

MJD is a fatal genetic neurodegenerative disease that develops in a similar way to MND, causing difficulties with movement and rendering patients immobile and dependent on their carers. Today, Laird's team investigates treatments for MJD and MND and she feels that these investigations complement each other.

*"... We can actually see the glowing neurons in the living animal."  
says Dr Angela Laird.*



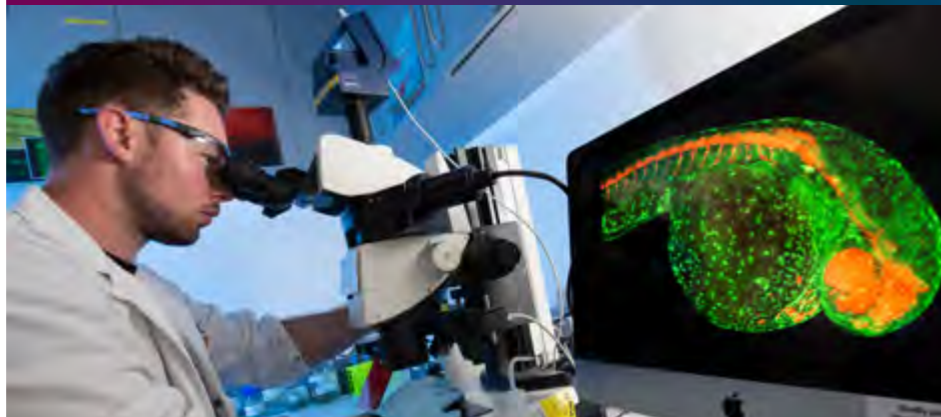
## FLUORESCENT FISH

An adult female zebrafish can breed every week, producing hundreds of eggs at a time. As the eggs are fertilised and develop outside the mother's body, they are perfect for the study of cellular processes, even in the living animal.

The embryos and juveniles of zebrafish are transparent, so Dr Laird's team is labelling their internal neuroanatomy with fluorescent markers.

"This means we can actually see the glowing neurons in the living animal. By looking down the microscope, we can determine which drug treatments are helping the neurons within zebrafish carrying MND genes to develop more normally. This can be done more rapidly than in other animal models," she says.

Zebrafish embryos also develop movement very early, within the first day after the egg is fertilised. "So that means for a disease like MND that affects movement, within a short timeframe we can see whether or not the mutated genes decrease their movement or whether drug treatments improve their movement," Laird says.



NEURODEGENERATION  
TREATMENT TEAM

Group Leader  
**Dr Angela Laird\***

Lecturer  
**Dr Emily Don\***

Postdoctoral Research Fellows  
**Dr Luan Luu\***  
**Dr Maxinne Watchon\***

Research Associate  
**Isabella Lambert-Smith**

Research Assistants  
**Caitlin Lucas\***  
**Katie Robinson\***  
**Claire Winnick**  
**Kristy Yuan\***

Masters Research Students  
**Maddy Tym**

\* Pictured

## NOVEL DRUGS AND NATURAL PRODUCTS

The team's investigation of potential treatments for MND includes testing novel drugs that have only just been synthesised and other drugs created to treat other diseases that could be repurposed to treat MND.

Her team is also starting to test natural compounds produced from plants and marine invertebrates through a collaboration with the Griffith Institute for Drug Discovery at Griffith University.

Successes so far include discovering positive effects of treating their zebrafish models of neurodegenerative disease with a few drugs that already have approval for human use from the Therapeutic Goods Administration.

These drugs remove some of the toxic proteins that cause those diseases. Testing of these drugs on the MND zebrafish is underway.

Laird's team is also collaborating with Professor Atkin's Cellular Neurobiology team to explore their potential drugs on the MND zebrafish, with some positive effects already seen.

The next step is to test the effect of the drugs that show the most potential on other preclinical models of MND, examining whether they can improve movement and protect against neurodegeneration in those models as well.





CELLULAR  
NEUROBIOLOGY TEAM

# Cell biology informs drug quest

Investigating cell biology and how MND operates at a molecular level is critical to informing the design of drugs to slow its progression and even cure the disease.

“We want to know how and why motor neurons die,” says Professor Julie Atkin, leader of the Cellular Neurobiology team, who is internationally recognised for her research on cellular mechanisms leading to neurodegeneration in MND.

*“So far, we’ve identified some cellular pathways that go wrong in MND and those are our biggest successes.”*

## DISEASE PROCESSES THAT CAUSE MND

Her team has found that transport of proteins within the motor neuron does not function properly in MND, and this is triggered by many different genes that cause genetic forms of the disease.

Some of the team’s discoveries include identifying that DNA damage is present in MND, and that the most important protein linked to MND, TDP 43, is a protein that normally repairs DNA. They’ve also identified another protein, C9orf72, that when mutated causes familial MND, commonly passed down

in families. This protein is normally required for cellular transport and in processes that cells use to get rid of damaged proteins.

Her team’s cellular investigation feeds back into their research on potential drugs. “The focus of our research is to pinpoint the first things that go wrong in MND, because then we can target drugs to prevent the disease progressing at the early stages,” says Atkin. “We’re trying to find the most upstream drug target because we believe this has the greatest probability of success.”



CELLULAR  
NEUROBIOLOGY TEAM

Group Leader  
Professor Julie Atkin\*

Postdoctoral Research Fellows  
Dr Mariana Brocardo\*  
Dr Eric Herbst  
Dr Cyril Jones Jagaraj\*  
Dr Anna Konopka  
Dr Sonam Parakh\*  
Dr Audrey Ragagnin  
Dr Hamideh Shahheydari  
Dr Marta Vidal\*

Research Officer  
Prachi Mehta\*

PhD Students  
Reka Toth  
Md Shafi Jamali\*  
Emma Perri (completed)  
Sina Shadfar\*

\* Pictured

## DRUG TESTING

Professor Atkin’s team is using this knowledge to test two types of new drugs, initially on cells grown in the lab.

An important tool in this research is a new high-content microscopy system, the first of its kind in Australia, which enables her team to examine and analyse cells very rapidly.

“It allows us to automate our analysis, which is a lot faster than doing it manually,” she says. Those drugs that prove effective in cells are then being trialled in zebrafish that develop MND, and then in transgenic models.

The team is also drawing on the expertise of chemists at La Trobe University in Melbourne and at the University of Queensland to improve the pharmacological properties of these drugs so they are more tolerable in the body.

“This is important for the drug to be effective on people, as well as stopping motor neurons from dying,” says Atkin.”

Hopefully, some of these drugs may eventually make it into human clinical trials.”

## TESTING HETEROGENEITY

The team recently received a new grant to use samples from the Neurodegenerative Disease Biobank to investigate MND’s heterogeneity – the different ways in which MND manifests itself in every patient. In some people, the disease presents in the limbs, but in others, in muscles that affect swallowing. Some people survive for years after diagnosis and others only weeks.

Importantly, some motor neuron cells live longer in MND than others. For example, motor neurons innervating the ocular muscles that control eye movement don’t degenerate until the very end stages of the disease, whereas motor neurons in the spinal cord fail early in the course of MND. This new project aims to determine why certain types of motor neurons are more susceptible than others.





MULTIDISCIPLINARY  
MND SERVICE AND CLINIC

# Clinic at the frontline in MND battle

The Multidisciplinary MND Service and Clinic, established in 2010, is the largest integrated MND clinic in Australia, managing 200 patients at any one time.

Located right next door to Macquarie University Hospital (MUH) and linked by an overhead walkway, the Multidisciplinary MND Service and Clinic was established to serve the multiple needs of MND patients. It currently manages about 10 per cent of people in Australia with the disease.

“It aims to be a one-stop shop,” says Professor Dominic Rowe, the neurologist who leads the Clinic. “It focuses on patient-centric care and we try to anticipate clinical issues before they arise.”

Patients come in every three months for consultations with a physiotherapist, speech pathologist, dietitian, occupational therapist,

social worker, clinical nurse consultant, genetic counsellor, respiratory physician and with Rowe, depending on their stage of the disease.

Costing \$500,000 a year to run, the Clinic operates entirely on donations, which cover salaries and access to care, no matter if the patient lives in Sydney or regionally. The Clinic supports remote and rural patients by telemedicine if required. The cost to the patient to visit the clinic is minimal (approximately \$30 out of pocket), with the actual cost met by donations.

Rowe and his team also look after MND patients in MUH. The Centre uses its

charitable fund to pay for patients without private health insurance to be admitted to hospital when appropriate.

All patients and their families are invited to participate in the Neurodegenerative Disease Biobank. The majority of patients have sporadic MND, but the Clinic currently cares for 62 families that have a faulty gene that causes their MND. There are now more than 20 genes known to cause inherited MND, and families have access to in-house genetic counselling at no cost.

Another goal is to establish low-cost access to advanced IVF techniques to ultimately eradicate the genes that cause MND.



*“We’ve had three babies born to our Clinic over the last four years where one of the parents has a faulty gene,” says Professor Dominic Rowe.*

*“With preimplantation genetic diagnosis and IVF, we know for certain that the babies do not carry the faulty gene and will never get MND. It’s the only instance in Australia and one of the few instances in the world where families have been cured of MND.”*



MULTIDISCIPLINARY MND  
SERVICE AND CLINIC STAFF

*Clinical Neurologist and Researcher*  
**Professor Dominic Rowe\***

*Clinical Nurse Consultant*  
**Kristina Barnes\***

*Clinical Trials Coordinator*  
**Richard Gan\***

*Speech Pathologist*  
**Sally Pittendrigh\***

*Dietitian*  
**Meaghan Joyce\***

*Occupational Therapist*  
**Kylie Christelz\***

*Physiotherapist*  
**Kate Bradbury\***

*Social Workers*  
**Elizabeth Hannan\***  
**Christie Charlin\***  
*Genetic Counsellors*  
**Rosie Fell\***  
**Ashley Crook\***

*Respiratory and Sleep Physician*  
**Corinna Pan\***

*Research Fellow*  
**Sinduya Subramanian\***

\* Pictured

## CLINICAL TRIALS

The Clinic is the springboard for the Centre’s clinical trials. One trial is a collaboration with the Albion Centre, the University of Sydney, Westmead Hospital and Calvary Health Care Bethlehem in Melbourne. Known as the Lighthouse Trial, it repurposed an antiretroviral therapy, called Triumeq.

The results of Phase 2 of this trial, which showed 88 per cent of patients stuck with the drug throughout the 24-week period, were published in July 2019. The drug was safe, well tolerated by patients and easy to administer. Initial results indicate Triumeq may have a positive biological effect, both clinically and on biomarkers, in patients with MND, perhaps slowing the progression of the disease in half the people taking it.

A much larger Phase 3 placebo-controlled trial will start in early 2020 to determine whether Triumeq improves survival and whether it is definitely able to slow clinical progression of people with MND. A total of 390 patients are participating at five sites in Australia, and others in Europe and the UK.

The MND Clinic is also the lead site of a world-first trial of a novel therapy called Copper ATSM. Phase 2 of this trial began in September 2019. Developed in Australia, Cu-ATSM aims to rebalance the metal deposition abnormalities that

occur in brains of MND patients, resulting in increased iron and reduced copper in neural tissue.

This novel medicine works by using a cellular transport mechanism to reduce the amount of iron deposited in neural tissue and increase the amount of copper. The initial Phase 1 study showed the medicine was safe and relatively well tolerated. The next phase will attempt to determine whether Cu-ATSM is able to slow the progression of MND.



**Costing \$500,000 a year to run, the Clinic operates entirely on donations.**



NEURO-IMPROVE GROUP

# Proteins unlock pathways to therapy

In MND, proteins aggregate and form large clumps inside nerve cells, which then die. Researchers are investigating why this happens.

Professor Roger Chung leads the NEURO-IMPROVE Group at the Centre for MND Research. IMPROVE stands for Imaging Proteomics Therapeutic Delivery. The group is composed of four interlinked teams that bring together diverse research expertise.

*“We’re interested in understanding why specific proteins stick together and how they cause MND,” says Chung, Associate Dean (Research) in the Faculty of Medicine and Health Sciences at Macquarie University. “We observe their actions in real time and ultimately use this information to develop novel therapeutics for MND.”*

## AN UNBIASED VIEW

Dr Albert Lee leads the Neuroproteomics team, which uses a range of experimental techniques to visualise protein-to-protein interactions, including mass spectrometry to look across all proteins, and a process called ‘unbiased proteomics’, to see how they behave.

Lee’s team seeks to understand how MND-specific protein aggregation alters normal protein pathways within cells and has identified some key proteins involved in the initial stages of MND-specific protein aggregation. They have discovered an important proteomic pathway significant to preventing MND-specific protein aggregation.

“This gives us an idea of how we can use this clearance pathway to our advantage and



stop MND-specific proteins accumulating and sticking together,” Lee says. The Centre has filed a portfolio of patents that describe this therapeutic concept and is performing preclinical therapeutic evaluation.

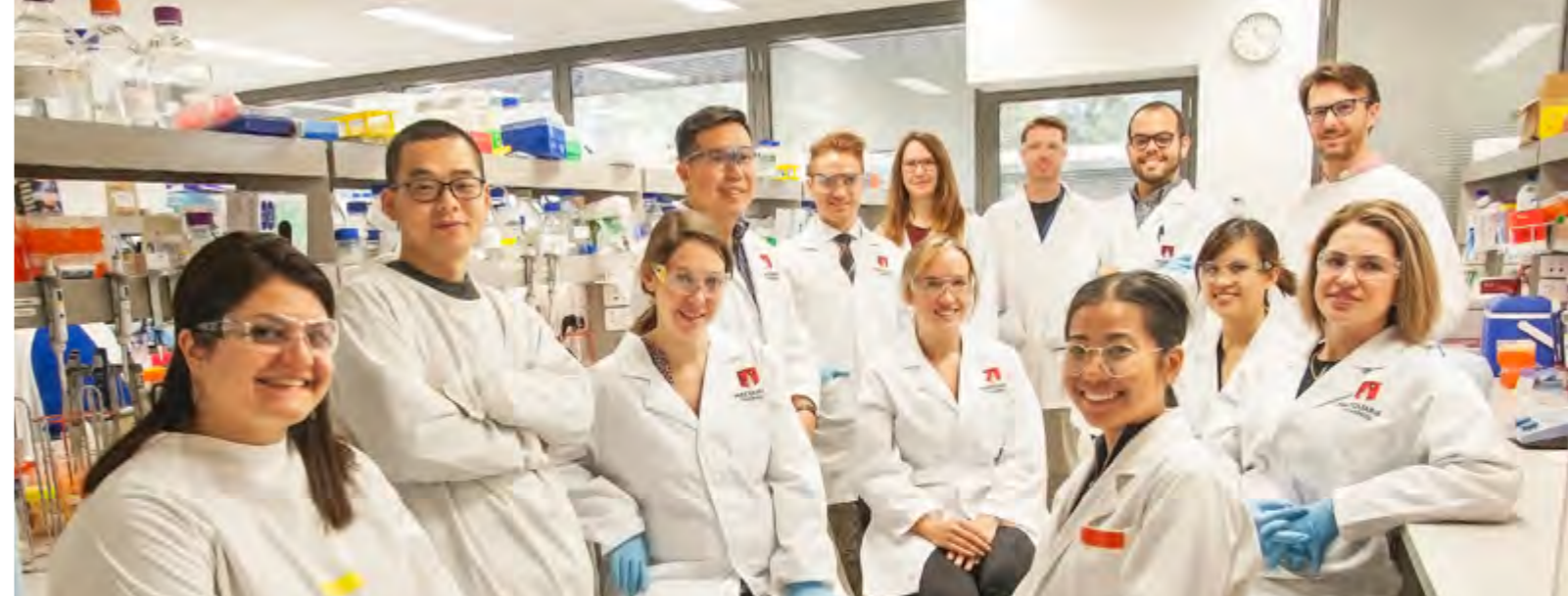
## FLUORESCENT MARKERS

Dr Marco Morsch’s Neuroimaging team works with zebrafish to visualise how MND-specific aggregates form and circulate through the nervous system. Zebrafish are transparent when they’re young, so researchers genetically modify fish to label their motor neurons and human MND proteins with fluorescent markers.

“This means we can watch how motor neurons respond to the disease in real time, and how the cells in the brain and spinal cord battle these disease aggregates,” says Morsch. “This provides a unique opportunity to observe how MND develops within nerve cells and how we can potentially target this therapeutically.”

Morsch’s team has identified a critical role of a particular immune cell during disease onset. These cells normally clear dying nerve cells and disease aggregates so that the disease proteins can’t spread to other cells.

“When we experimentally interfered with this ‘housekeeping service’, we discovered that the aggregates can disperse in unprecedented ways. Once we understand these pathways better we hope to identify novel opportunities to delay disease progression,” Morsch says.



## DELIVERING DRUGS WITH NANOPARTICLES

A significant hurdle for any therapeutic treatment for neurodegenerative diseases is how to deliver drugs to the brain or spinal cord. They are covered with a protective barrier called the blood-brain barrier, a layer of cells specifically designed to keep the brain separate from the rest of the body. A second challenge once drugs cross this barrier is how they can specifically target diseased cells and not healthy ones.

Dr Bingyang Shi’s Brain Drug Delivery team is developing ‘smart’ nanoparticle-based solutions to carry the drugs into the brain and target specific cells. In one method, he puts the drugs in a glucose-coated nanoparticle that is readily transported

into the brain and nerve cells, because nerve cells in the brain have a high energy requirement (they generate it from glucose in the diet).

## DIAGNOSIS AND PROGNOSIS

Dr Chung’s Biomarkers and Disease Mechanism team is identifying new disease biomarkers using unbiased molecular technologies that may allow earlier disease diagnosis or measures of disease progression and severity.

“We are looking to identify precisely how things go wrong inside motor neurons in MND, which will give us new ideas for how we can protect the neurons and find out whether there are biomarkers in the blood that can inform us of disease onset or progression,” Chung says.



NEURO-IMPROVE GROUP MEMBERS

Group Leader  
**Professor Roger Chung\***

Senior Lecturer, Team Leader  
**Dr Albert Lee\***

Senior Research Fellow, Team Leader  
**Dr Marco Morsch\***

NHMRC Dementia Research Fellow, Team Leader  
**Dr Bingyang Shi\***

Research Associate  
**Stephanie Rayner\***

Research Assistants  
**Flora Cheng**  
**Rowan Radford\***

Postdoctoral Research Fellows

**Dr Libing Fu**  
**Dr Jia Li**  
**Dr Cindy Maurel**  
**Dr Amanda Wright\***  
**Dr Anne Zhou**

PhD Students  
**Afshin Babazadeh\***  
**Jennilee Davidson\***  
**Natalie Scherer\***  
**Hannah Suddull\***  
**Andres Vidal-Itriago\***  
**Guoying Wang\***

Masters Research Students  
**Bahar Kavyani\***  
**Paulina Szwaja**

\* Pictured



# Kaitlin, Jessica and Susan

In 2010, Kaitlin and Jessica Ellis' uncle lost his battle with motor neuron disease. Five years later, their mother, Susan, started showing similar symptoms. Her foot began dropping and she frequently fell over. Doctors thought she had arthritis.

Kaitlin and Jessica's worst fears were realised when their 57-year-old mother tested positive for MND. They immediately gave up their jobs and study to jointly care for Susan full-time at home for two-and-a-half years. Susan died in 2017.

"It was so hard to watch our mum, knowing she had a terminal illness with no cure. The person we loved the most just declined more and more every day," says Kaitlin, who was 21 at the time and working as a nurse. "Mum was everything to us. We looked after her 24/7 and

faced many challenges, but it brought us all much closer together." Jessica, 24 at the time, was a legal secretary.

Susan's doctors at Liverpool Hospital suggested her daughters undergo genetic testing. They discovered they both carried the gene that caused MND. "Getting genetic testing is a very personal choice and everyone reacts differently," Kaitlin says. "I'd seen how my uncle and Mum had suffered and I didn't want anyone else in my family to suffer like that, let alone myself."

Jessica and Kaitlin chose to get genetic testing so that in future, they can go through IVF and hopefully have a child who isn't a carrier. The sisters now give annual blood samples to the Neurodegenerative Disease Biobank.

"In retrospect, I wish I had waited until after Mum had passed away to get tested," Kaitlin says. "When I told her, it absolutely broke her. She cried and cried. It was devastating for all of us."



The sisters have set up a support group called MND Genies, for people like themselves who have an MND genetic mutation but haven't yet developed the disease.

# Cobie, Luke and Mick

Luke Smith's father, Mick died from MND in 2017. He'd been diagnosed about 15 years earlier. Luke's two siblings had genetic testing and had no trace of the genes known to cause MND. When Luke took the test, he discovered he was a carrier of familial MND.

"It took a long time to sink in and I felt pretty horrible," says Luke. "But I came good after about six months." Newly married to Cobie, and the couple, both 24 years old at the time, decided to use IVF to conceive.

"It was pretty clear to us once I discovered I was gene-positive," Luke says. "We'd watched my father deteriorate and suffer. We knew we wanted to have kids but we didn't want them to carry this gene, so doing IVF was an easy decision for us. The technology's there, so why not use it?"

Luke and Cobie started the IVF process in March 2012. Cobie says it was very difficult and stressful. "I found it hard mentally and I

wouldn't wish it on my worst enemy," she says. "But it has given us peace of mind down the track."

The couple was fortunate to have financial backing but would have loved to have more emotional support. "We were the first couple with hereditary MND that our fertility doctor had dealt with," Cobie says. "I felt totally isolated. There was no one else in my life who could understand what I was going through. We were walking into the unknown."

Their first daughter was born in December 2013 and their second in 2017. Cobie says she'd love to talk to other couples to give them the support they themselves never had.



Luke provides biological samples to the Neurodegenerative Diseases Biobank. Mick was a patient of Professor Dominic Rowe at the Multidisciplinary MND Service and Clinic.

## GENETIC COUNSELLING SERVICE

Rosie Fell is one of the genetic counsellors at the Multidisciplinary MND Service and Clinic. She counsels patients and relatives of patients about familial MND. Individuals may be referred to her by Professor Dominic Rowe if he suspects they have inherited MND because multiple members of their family have had the disease. Some individuals request a referral themselves, as they are concerned about a personal or familial risk of MND.

Fell gathers family history information from both maternal and paternal sides. If relevant, she will then offer genetic testing. "Our role is to give our patients all the information in a way that they can understand, so they can make an informed decision about their health and decide what to do next," Fell says. "If their MND is genetic, then that may have implications for other family members."

# Giving for future generations

A few years ago, Mike Chisholm's mother died of MND in New Zealand. Since then, he's become a regular financial donor to the Centre for MND Research because he's impressed by the passion and commitment of the researchers and clinicians.

When Beth Chisholm died of MND in New Zealand in 2011, her son, Mike, who was living in Sydney and CEO of Crestone Wealth Management, took a keen interest in investigating MND research around the world. He soon found the Centre for MND Research at Macquarie University.

"I took the initiative to go out to the Centre and see what they were doing," Mike says. "I found a group of extremely talented but equally passionate people. For them, it is far more than just a job. They are fully invested in what they do and that's why they've had such incredible results."

Mike was really amazed by the Centre's multidisciplinary approach, providing tailored care for MND patients and their families,

while researchers also conduct ground-breaking research and world-first clinical trials. "I am extraordinarily impressed with what they've created from nothing."

In 2016, Mike became an annual donor to the Centre and also actively participates in the Centre's gala and fundraising events. He encourages other people who can, to donate.

"If people are lucky enough to have either well-paying jobs or have inherited reasonable sums of money or gained it through some other event, then I have a view they have a responsibility to support the community we live in," Mike says.

Giving to an organisation such as the Centre for MND Research helps develop treatments and cures for future generations, he says. "I also get a huge satisfaction, a sense of pride, when I hear what the team at the Centre has been able to achieve, their discoveries and ongoing research. They're solving really challenging problems and are going to have a huge impact on a large number of lives and families across Australia and the world."



# Alumni urged to donate

Alumnus David Bussman gives back to his Alma Mater and encourages others to give to the Centre that relies on private donors and research grants.



After retiring from a career as a Chartered Accountant, David Bussman decided to pursue his passion in Egyptology and enrolled in a Master of Arts at Macquarie in 1996. "The promise of a study trip to Egypt for four weeks which included inspecting Macquarie digs added to the attraction," he says.

David continued his connection with Macquarie, and with his wife Patricia, decided to invest in future generations by leaving the University a bequest in their will. They chose the Centre for MND Research after a lab tour.

"The researchers had impressive zeal and optimism, and realised they may well be 'in the trenches' for a long haul before they can win the fight against MND," says David. He believes in 'leading from the front' and hopes other alumni and employees will follow suit.

# Country town rallies

Residents of Bathurst, three hours' drive west of Sydney and best-known for its annual 1000km supercar race, showed their generous spirit when a local developed MND.

Tony Campbell, a businessman in Bathurst, was diagnosed with MND in March 2018. His decline was swift, says his partner Janine Graham. And when the couple visited the Centre for MND Research and attending Professor Dominic Rowe's Multidisciplinary MND Service and Clinic, they decided to raise funds to support its work.

"Seeing all the younger people who'd been diagnosed with MND made us really want to do something," Janine says. "It's too late for Tony, but maybe in the next few years researchers will find what causes this disease or find a cure."

Janine began organising fundraising efforts and in April 2019, local townspeople in

Bathurst showed their overwhelming support, donating more than \$50,000.

"Country towns are very good at rallying around one of their own who is going through difficult times," Janine says. "It was really fun for Tony and I to organise the night and it gave us something different to focus on a while."

# Events

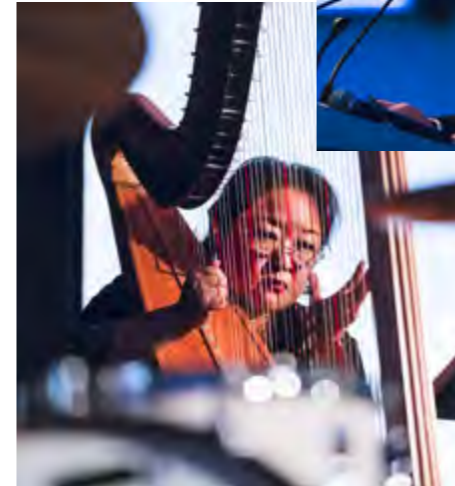
## FIRIES CLIMB FOR MND

In 2015, Matthew Pridham approached the Centre for MND Research after his best friend, Adam Regal, was diagnosed with MND. Pridham, a NSW firefighter, wanted to make a difference. This was the genesis of an annual fundraising event held every spring, when sponsored firefighters climb the 1,504 steps of the Sydney Tower Eye wearing 20kg of gear. So far, they've raised more than \$2 million, all of which supports MND research and clinical trials. Adam Regal died in 2018. The event in his memory helps support other Australians living with MND and their families.



## ANNUAL CONFERENCE

The Centre also hosts the annual Macquarie Neurodegeneration Meeting, a national conference attended by more than 150 researchers. The conference showcases the latest research on MND, frontotemporal dementia, Alzheimer's, Parkinson's diseases, and other degenerative brain disorders, and provides an opportunity for Australian neuroscientists to network and collaborate on further research.



## GALA

Every year, the Centre for MND Research and the Multidisciplinary MND Service and Clinic hold a gala fundraising dinner and auction — 2019 was no exception. On 28th June, 450 supporters attended the event at the Four Seasons Hotel in Sydney. Professor Dominic Rowe presented an overview of the Centre and Clinic, highlighting patient care, research, clinical trials and therapies.

This year, the event raised more than \$215,000 through a silent auction, which included Jervis Bay and Hunter Valley holidays and a Ken Done original painting. The live auction items included MND lab tours and a purebred Poll Hereford Heifer, while the famous 10-person lunch in Bulga, prepared and served by Rowe, raised \$10,000 in its own right.

## LAB AND CLINIC TOUR

Experience first-hand what keeps researchers and clinicians busy at the Centre for MND Research and the Multidisciplinary MND Service and Clinic. Visitors receive a guided tour of the Clinic, where more than 10 per cent of Australia's MND patients are cared for by Professor Dominic Rowe and his team, and where two Australian-first clinical trials are in progress. They go behind the scenes into the laboratories and meet the outstanding research teams tackling MND, from diagnosis to treatment, in their quest for a cure.



# Community funds are vital for future work

Fundraising and community support are essential to ensuring the Centre for MND Research and the Multidisciplinary Service and Clinic can continue to improve the outcome of people living with MND.

We are so grateful to the thousands of people, organisations, communities and foundations that continue to support our work.

Since 2013, the Centre, Service and Clinic have received almost \$9 million in philanthropic support from our community of donors. It has helped form the most comprehensive and all-encompassing MND research centre in Australia, where researchers and clinicians work side by side to decipher this disease.

No other centre has this kind of firepower to deliver the science and breakthroughs that will disrupt the trajectory of MND. The Service and Clinic receive no federal or state funding to operate. They are run entirely with the support of public donations and research grants. At the Centre, our researchers compete for grants from the National Health and Medical Research Council, the Australian Research Council and the Motor Neurone Disease Research Institute Australia, among others.

It is only with adequately funded research that

we can uncover how to beat MND. We know it can be done – but we need your help to make it happen. Your donations support a variety of research and clinical initiatives, including:

- identifying genetic, lifestyle and environmental risk factors to help understand why people develop MND and prevent its onset and progression
- new drug discovery and development – supporting innovative strategies to identify potential new therapies
- clinical trials – allowing us to test new therapies for MND, which may slow and stop the disease
- biomarker discovery and development – tracking progression and monitoring the effectiveness of potential new drugs
- Neurodegenerative Disease Biobank – funding the continual collection, analysis and preservation of biological samples, which help us understand the causes and biomarkers of MND

- early and mid-career fellowships – helping us recruit and support the best researchers to unravel the mysteries of MND
- Multidisciplinary MND Service and Clinic – giving patients access to best practice subsidised MND care.

If you would like to find out how you can support the Centre for MND Research and the Multidisciplinary MND Service and Clinic, please contact Macquarie University's Advancement team:

**T:** 1800 673 662

**E:** [mqadvancement@mq.edu.au](mailto:mqadvancement@mq.edu.au)

Or visit

**[mq.edu.au/mnd](http://mq.edu.au/mnd)**

to make a donation online.

**Thank you for making a difference.**



# Our Leadership Team



**PROFESSOR IAN BLAIR**  
Centre Co-Director and Group Leader

Professor Ian Blair has a long-standing track record in research identifying and understanding the genetic and genomic factors that underlie the onset and progression of motor neuron disease (MND/ALS) and related disorders.

**Contact:** [ian.blair@mq.edu.au](mailto:ian.blair@mq.edu.au)

**PROFESSOR DOMINIC ROWE AM**  
Centre Deputy Director

Professor Dominic Rowe's areas of expertise include MND and Parkinson's disease among other forms of neurodegenerative disease. As the inaugural Professor of Neurology at Macquarie University, he also researches the causes of and therapies for these diseases.

**Contact:** [dominic.rowe@mq.edu.au](mailto:dominic.rowe@mq.edu.au)

**PROFESSOR JULIE ATKIN**  
Centre Co-Director and Group Leader

Professor Julie Atkin is interested in the basic molecular and cellular mechanisms in MND. She is developing new therapeutic strategies to prevent neurodegeneration. Her work focuses on the role of DNA damage, cellular stress responses, redox signalling and cellular trafficking pathways.

**Contact:** [julie.atkin@mq.edu.au](mailto:julie.atkin@mq.edu.au)



**PROFESSOR GILLES GUILLEMIN**  
Centre Executive Member and Group Leader

Professor Gilles Guillemain is internationally recognised as a leader in the fields of tryptophan, neuroinflammation and neurotoxicity research. His team has three main interests:

- identification of environmental factors associated with MND
- developing new sets of blood biomarkers for MND progression and response to treatment
- developing therapeutic strategies for MND using new enzyme inhibitors.

**Contact:** [gilles.guillemain@mq.edu.au](mailto:gilles.guillemain@mq.edu.au)

**DR ANGELA LAIRD**  
Centre Executive Member and Group Leader

Dr Angela Laird studies the pathogenesis of movement disorders, with a particular focus on identifying and testing potential disease treatments. Her group is currently performing drug-testing studies on zebrafish and cellular models of MND and related diseases. The best therapeutic candidates will receive further preclinical investigation to aid development of effective treatments for MND.

**Contact:** [angela.laird@mq.edu.au](mailto:angela.laird@mq.edu.au)

**PROFESSOR ROGER CHUNG**  
Centre Executive Member and Group Leader

Professor Roger Chung's team explores the neurobiology and neurochemical pathways that underlie MND. His team has four main interests:

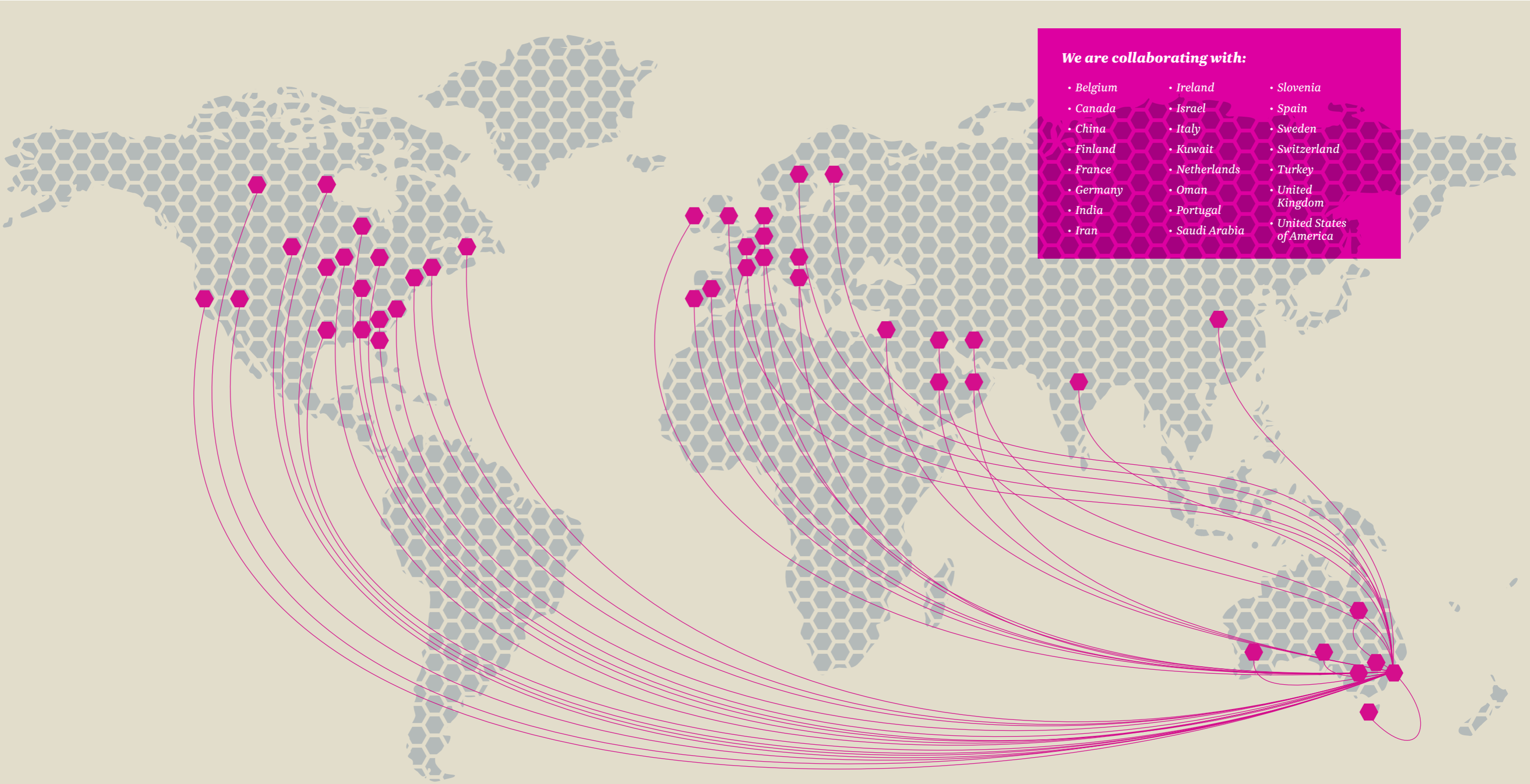
- molecular and proteomic understanding of the origins of MND
- understanding how non-neuronal cells contribute to disease processes
- developing biomarker assays for disease onset and progression
- developing therapeutic strategies for MND using gene therapies and nanoparticle-based drug delivery systems.

**Contact:** [roger.chung@mq.edu.au](mailto:roger.chung@mq.edu.au)



# Global institutional collaborators

The Centre for MND Research has collaborated with 423 research institutions from all over the world.



- We are collaborating with:**
- Belgium
  - Canada
  - China
  - Finland
  - France
  - Germany
  - India
  - Iran
  - Ireland
  - Israel
  - Italy
  - Kuwait
  - Netherlands
  - Oman
  - Portugal
  - Saudi Arabia
  - Slovenia
  - Spain
  - Sweden
  - Switzerland
  - Turkey
  - United Kingdom
  - United States of America

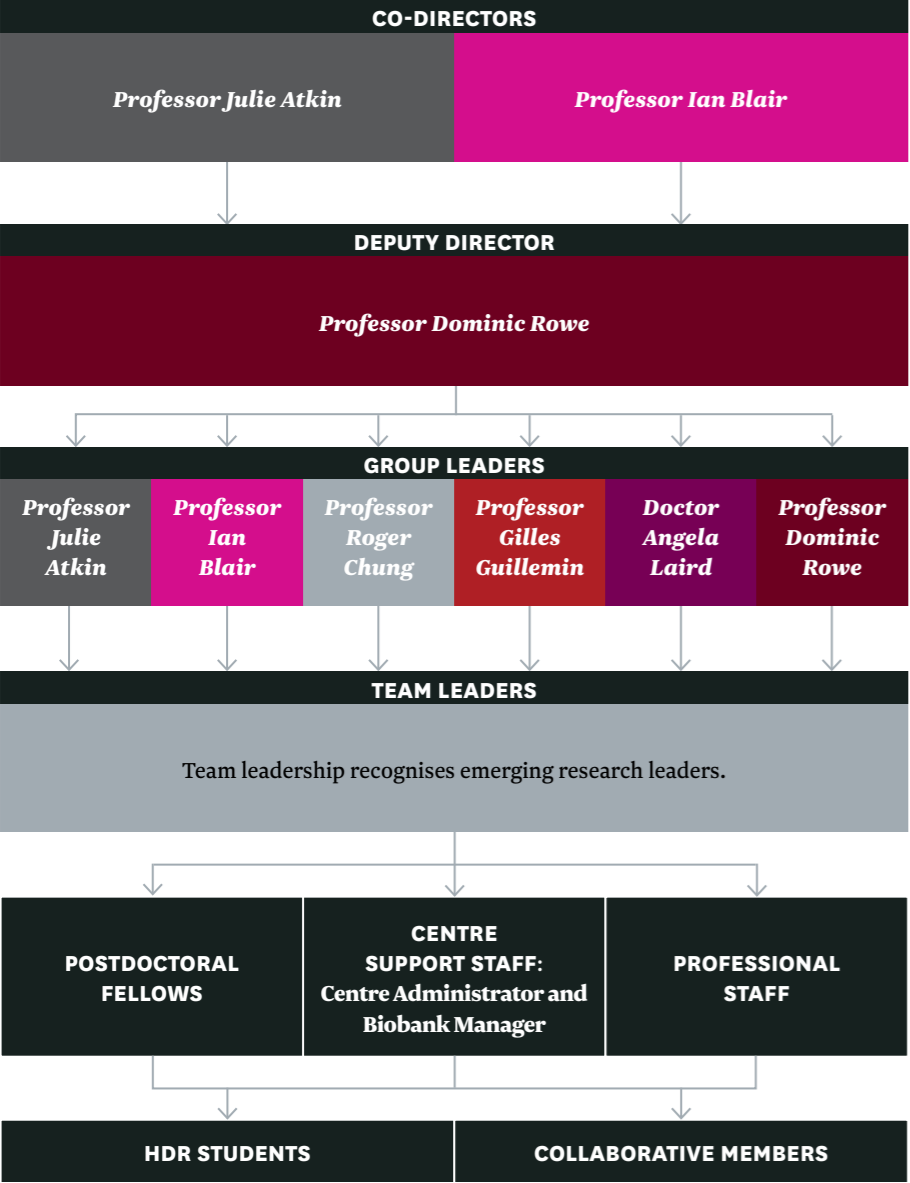
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# Governance structure

## Centre leadership



### DIVERSITY

- 1 | 1** We have one female and one male co-director of the Centre.
- 19 | 8** We have 19 female research fellows and eight male research fellows.
- 2 : 4** Our current female to male Group Leader ratio is 2:4.
- 60%** Approximately 60% of our HDR students are international, providing a diverse and stimulating environment in the Centre.

### GOVERNANCE

The key Centre leadership group is the Executive. Operational committees responsible for specific activities include the Biobank Committee, Philanthropy Committee, Senior Scientist Group, Conference Working Group and Laboratory Operations Committee.

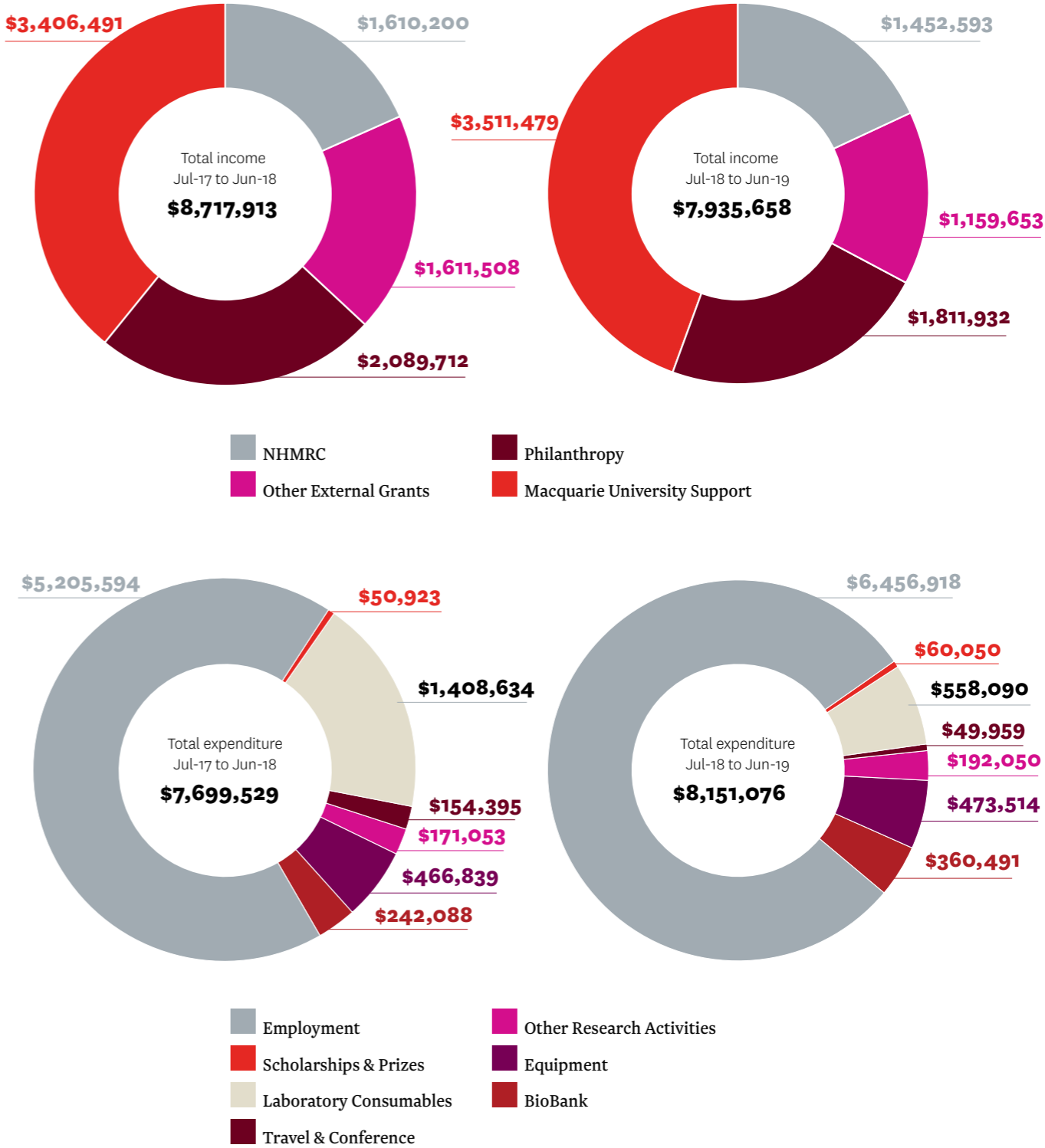
### EXECUTIVE GROUP

- Centre Directors
- Deputy Director
- Group leaders
- One representative from each academic level (A-D), drawn from the Centre (by nomination for 12-month term)
- One HDR student (by nomination for 12-month term)
- Centre Administrator

The Executive Group meets in person every two months, chaired by a Centre Director, to set strategic direction for the Centre and oversee operational management through the various committees listed above. The Executive is responsible for managing the Centre budget including philanthropic funds, as well as intellectual property, ethics approvals and contracts, upon the advice of respective Committees.

# Financial highlights

Summaries of the income and expenditure, in 2017-18 and 2018-19, for the Centre for MND Research are illustrated below. We are immensely grateful for the philanthropic support, which is critical for underwriting scientists and sustaining the Centre’s ongoing research.



# Major grant highlights 2018/2019

Our researchers at the Centre for MND Research receive highly competitive grants that support their salaries and research programs. Highlighted grants new and ongoing, including those from the National

Health and Medical Research Council of Australia (NHMRC), are listed below. Our outstanding grant success reflects the recognition of our peers for our highly novel and innovative research programs.

Funding Body	Type of Grant	Principal Investigator	Co-Investigators	Project Title	Amount Funded	Years Funding
Motor Neurone Disease Research Institute of Australia inc	MNDRIA Innovator Grant	Roger Chung	Albert Lee; Stephanie Rayner; Shu Yang	Revealing the role of protein clearance pathways in sporadic ALS	\$100,000.00	1
Motor Neurone Disease Research Institute of Australia inc	MNDRIA Betty Laidlaw MND Research Prize 2019	Marco Morsch	Albert Lee; Adam Keith Walker; Roger Chung	Targeting the nucleo-cytoplasmic transport machinery in sporadic and familial ALS	\$250,000.00	1
National Health and Medical Research Council	NHMRC Early Career Fellowship	Kelly Williams		Unravelling the molecular basis of amyotrophic lateral sclerosis	\$334,436.00	6
FightMND	IMPACT Grant	Julie Atkin	Ian Blair; Dominic Rowe; Edwin Lim	Disease susceptibility, precision medicine and ALS	\$249,742.00	3
FightMND	Drug Development Grant	Julie Atkin	Adam Keith Walker; Angela Laird; Begona Heras (external); Marilyn Anderson (external); David Craik (external); Mark Bleackley (external)	Therapeutics based on the protective activity of protein disulphide in MND (PDI project)	\$1,208,147.00	4
FightMND	Drug Development Grant	Julie Atkin	Adam Keith Walker; Angela Laird; Baell Jonathan (external); Belinda Abbott (external)	Novel therapeutics targeting neuronal transport pathways in MND (Rab1 project)	\$1,205,035.00	4
Motor Neurone Disease Research Institute of Australia inc	MNDRIA Research Grants	Jenn Fifita	Kelly Williams; Ian Blair	Investigating the role of large structural variation in MND by analysis of whole-genome sequencing data	\$100,000.00	2
Motor Neurone Disease Research Institute of Australia inc	MNDRIA Bill Gole MND Postdoctoral Fellowship	Jenn Fifita	Ian Blair	Investigating the molecular and pathological basis of Amyotrophic Lateral Sclerosis (ALS)	\$375,000.00	4
National Health and Medical Research Council	NHMRC Dementia Research Team Grants	Ian Blair	Julie Atkin; Roger Chung; Gilles Guillemain; Lezanne Ooi (external); Bauer Denis (external); Mark Molloy; Justin Yerbury (external); Nicholas Cole; Angela Laird; Tim Karl (external); William Wilson (external)	Developing insight into the molecular origins of familial and sporadic frontotemporal dementia and amyotrophic lateral sclerosis	\$7,277,279.00	5
National Health and Medical Research Council	NHMRC - Investigator Grants	Ian Blair		Determining the molecular basis of amyotrophic lateral sclerosis	\$2,198,640.00	5
National Health and Medical Research Council	NHMRC - Investigator Grants	Gilles Guillemain		The Kynurenine Pathway in neuroinflammatory diseases: translation to prognostics and therapeutics	\$2,401,595.00	5
Motor Neurone Disease Research Institute of Australia inc	MNDRIA Robert Turnbull MND Research Grant	Kelly Williams	Lyndal Henden; Ian Blair	Comprehensive transcriptome analysis of neuroanatomical regions with variable pTDP-43 pathology in sporadic ALS patients	\$100,000.00	1
Motor Neurone Disease Research Institute of Australia inc	MNDRIA Research Grant	Albert Lee	Stephanie Rayner; Adam Martin; Annika van Hummel; Marco Morsch; Bingyang Shi; Yazi Ke; Lars Ittner; Roger Chung	Clearance of TDP-43 by PROteolysis Targeting Chimera (PROTAC) dual targeting to treat amyotrophic lateral sclerosis (ALS)	\$100,000.00	1
Motor Neurone Disease Research Institute of Australia inc	MNDRIA MonStAR MND Research Grant	Marco Morsch	Cindy Maurel; Albert Lee; Patrick Vourc'h; Roger Chung	The unexplored posttranslational modification (SUMOylation) of TDP43 affects aggregate formation and localisation	\$100,000.00	1
Motor Neurone Disease Research Institute of Australia inc	Peter Stearne Familial MND Research Grant	Julie Atkin	Thomas Fath	Novel mechanisms of neurodegeneration induced by dysfunctional actin dynamics in MND	\$99,324.00	1

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To make a donation, visit

[mq.edu.au/mnd](http://mq.edu.au/mnd)

**We all work together  
with the one goal –  
to stop MND.**

**We know it can be done,  
we just need your help to  
make it happen.**

