Australian Government
Australian Research Council

EI 2018 ENGAGEMENT AND IMPACT

Engagement and Impact 2018

Macquarie University

MQU11-BCS (HLS) - Impact

Overview

Title

(Title of the impact study)

Treating Motor Neuron Disease

Unit of Assessment

11 - Medical and Health Sciences

Additional FoR codes

(Identify up to two additional two-digit FoRs that relate to the overall content of the impact study.)

06 - Biological Sciences

Socio-Economic Objective (SEO) Codes

(Choose from the list of two-digit SEO codes that are relevant to the impact study.)

92 - Health

Australian and New Zealand Standard Industrial Classification (ANZSIC) Codes

(Choose from the list of two-digit ANZSIC codes that are relevant to the impact study.)

84 - Hospitals
85 - Medical and Other Health Care Services

Keywords

(List up to 10 keywords related to the impact described in Part A.)

Health

Medicine

Motor Neurone Disease

Genetics

Biobanking

Academic Health-Science Centre

Integrated Health Care

Clinical Trials

Community Philanthropy

Sensitivities

Commercially sensitive

No

Culturally sensitive

No

Sensitivities description

(Please describe any sensitivities in relation to the impact study that need to be considered, including any particular instructions for ARC staff or assessors, or for the impact study to be made publicly available after EI 2018.)

Aboriginal and Torres Strait Islander research flag

(Is this impact study associated with Aboriginal and Torres Strait Islander content?
NOTE - institutions may identify impact studies where the impact, associated research and/or approach to impact relates to Aboriginal and Torres Strait Islander peoples, nations, communities, language, place, culture and knowledges and/or is undertaken with Aboriginal and Torres Strait Islander peoples, nations, and/or communities.)

No

Science and Research Priorities

(Does this impact study fall within one or more of the Science and Research Priorities?)

Yes

Science and Research Priority	Practical Research Challenge
Health	Better models of health care and services that improve outcomes, reduce disparities for disadvantaged and vulnerable groups, increase efficiency and provide greater value for a given expenditure.

Impact

Summary of the impact

(Briefly describe the specific impact in simple, clear English. This will enable the general community to understand the impact of the research.)

Macquarie Neurology and the MND Service at Macquarie University runs the largest specialist clinic for patients with Motor Neuron Disease (MND). The co-location of academic clinical services with Australia's largest concentration of MND researchers provides patients with bespoke, patient-focussed care. This supports a translational research platform that has made important discoveries of genetic origins of familial MND, which has enabled delivery of MND pre-implantation IVF services to give birth to babies free of MND in Australia. Our genetic discoveries have also been incorporated into standard practice diagnostic genetic testing of MND patients that are used worldwide.

Beneficiaries

(List up to 10 beneficiaries related to the impact study)

Patients

Community

Clinicians

Government Health Departments

Biobank Users

Countries in which the impact occurred

(Search the list of countries and add as many as relate to the location of the impact)

Australia

Details of the impact

(Provide a narrative that clearly outlines the research impact. The narrative should explain the relationship between the associated research and the impact. It should also identify the contribution the research has made beyond academia, including:
- who or what has benefitted from the results of the research (this should identify relevant research end-users, or beneficiaries from industry, the community, government, wider public etc.)
- the nature or type of impact and how the research made a social, economic, cultural, and/or environmental impact
- the extent of the impact (with specific references to appropriate evidence, such as cost-benefit-analysis, quantity of those affected, reported benefits etc.)
- the dates and time period in which the impact occurred.

NOTE - the narrative must describe only impact that has occurred within the reference period, and must not make *aspirational claims.)*

GENETIC TESTING AND PATIENT IMPACT

The availability of genetic testing is particularly important for patients with inherited forms of MND. Prof Blair and his team have collected DNA from over 200 Australian families with family history of MND, and have now mapped the causative gene mutation in about 60% of these families. These research discoveries have an important impact that Macquarie provides for our patients through the provision of the first and only genetic counsellor employed within a MND clinic in Australia. This provides expert advice and genetic counselling for patients and their families. A topic of common concern for patients is the risk that they may pass MND onto their children (most causative mutations are dominantly inherited, therefore 50% chance of offspring developing MND). Until recently, the options for parents were to either not have children, or “take the risk”. To address this, the Macquarie MND clinic has worked to provide the first and only MND pre-implantation genetic IVF service in Australia, delivered through Genea. To date, we have delivered three babies born “MND free”. This service was made possible by Macquarie’s genetic discoveries and translation of those discoveries into tools for parents to ensure their children do not inherit MND-related gene mutations.

THE MACQUARIE MODEL FOR MND CLINICAL CARE

For more than 10 years, Professor Dominic Rowe has led the MND clinic based at Macquarie University. He offers multidisciplinary care for MND patients in an out patient setting, where in a single session the patient has access to a group of MND-specific medical experts including neurologist, clinical nurse consultant, speech pathologist, orthotics, respiratory support, social worker support, physiotherapist, dietician, etc – this delivers integrated patient care that improves the quality of healthcare provided to MND patients. It is also delivered from a single location on the same day, and prevents patients and their families having to co-ordinate these medical services from multiple providers at different sites, which can be complicated, stressful and expensive. Notably, the multidisciplinary MND clinic is offered at little cost to patients, with all costs outside of rebatable support covered by donations to Macquarie University. The financial “gap” is approximately $400,000 per year, and therefore Macquarie is making a positive and highly appreciated impact for patients.

The integrated nature of the MND clinic and Macquarie University Hospital is also important. As the disease progresses, MND patients might be hospitalised. There is full integration of clinical care for MND patients between both clinic and hospital, with specialist MND nurses in the hospital who provide care in direct guidance from the consulting MND neurologist (who is able to regularly check on patients because of the direct proximity). For MND this is critical, analysis of the outcomes for MND patients across Australia has shown that this care model creates a statistically significant increase in the length of a patient’s life. For a terminal illness with no current cure this is a major impact on the patient and their families. In addition, the MND Service at Macquarie University has an indigent fund that pays for the care of patients in Macquarie University Hospital who might not have private health insurance.

The professional and personalised care that is provided to MND patients through the MND clinic has translated into strong patient support for the Macquarie MND biobank that facilitates research activities. Built upon our longstanding experience in the collection of DNA samples over 20 years for genetic research, over the past three years we have established the largest national MND biobank collecting longitudinal samples from patients each time they visit the clinic (generally every 3-6 months). We currently collect blood samples (for genetic testing, blood biomarker discovery research), urine samples, hair samples, and for some patients we collect skin biopsies for induced pluripotent stem cell generation. The strong rapport of patients and their families with the clinical team facilitates most of them providing ongoing samples into the biobank. To date, we have more than 600 participants in the MND biobank (roughly 400 patients, 200 controls). However, we do not view this simply as a transactional relationship. This is evidenced through our commitment to patients first, through delivery of the multidisciplinary clinic and by actively engaging patients in our research activities. Patients and their families are taken on tours of the University’s MND research facilities – located one floor below the clinic. We show them how their generous contribution to the biobank enables research towards finding a treatment for MND. We also have researchers visit the multidisciplinary clinic as observers, to embed a holistic integration of the MND research pipeline and the critical importance of patients within it. A strong indication of the impact that this has upon patients is through their willingness to continue providing ongoing samples to the biobank. In turn, this ensures that we have a rich biobank comprised of longitudinal samples collected across the disease timecourse, which will ultimately be a key resource for MND research.

IMPACT UPON COMMUNITY

MND has a devastating impact upon the Australian community. The way in which the disease manifests and progresses is emotionally distressing for families and friends to observe. While it has publically been considered a rare disease, latest national data indicates that two Australians die from MND each day – it is not rare. A recent economic analysis undertaken by KPMG under the commission of MND Australia identified that the financial cost of care per patient per year is approximately $1million. Therefore, this is an extremely costly disease (socially and economically) to the Australian community.

Associated research

(Briefly describe the research that led to the impact presented for the UoA. The research must meet the definition of research in Section 1.9 of the EI 2018 Submission Guidelines. The description should include details of:
- what was researched
- when the research occurred
- who conducted the research and what is the association with the institution)

The Macquarie University Centre for MND Research is the home of Australia’s largest clinical and research program in MND. The Centre seeks to understand the molecular origins of MND and develop new treatments for evaluation through clinical trials. This multidisciplinary research centre was established together with six research teams to collectively form a research pipeline that connects the MND clinic to human genetics, cell biology and biochemistry, animal models of MND, and pre-clinical testing. The goal is to make breakthrough discoveries in understanding the causes of MND and identify and evaluate new diagnostic and therapeutic strategies through preclinical and clinical trials. The colocation of MND research leaders across multiple disciplines forms a research ecosystem to maximise translational opportunities as well as national and international collaboration. For example, the Macquarie Centre leads Australia’s role in Project MinE, the world’s largest genetic study in MND. The MND Service has embedded genetic cousellors, who are critical to the adroit and ethical handling of genetic information and the process of moving forward into genetic testing. This is also a fertile area for research as understanding the elements that are involved in genetic testing are crucial to the process of informed consent. Health economics will also inform the process of genetic testing in the setting of pre implantation genetic diagnosis with assisted fertilisation techniques.

FoR of associated research

(Up to three two-digit FoRs that best describe the associated research)

06 - Biological Sciences
11 - Medical and Health Sciences

References (up to 10 references, 350 characters per reference)

(This section should include a list of up to 10 of the most relevant research outputs associated with the impact)

NEK1 variants confer susceptibility to amyotrophic lateral sclerosis.
Kenna KP, et al. Nat Genet. 2016 Sep;48(9):1037-42

CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia.
Williams KL, et al. Nat Commun. 2016 Apr 15;7:11253.

Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways.
Cirulli ET, et al. Science. 2015 Mar 27;347(6229):1436-41.

Exome-wide rare variant analysis identifies TUBA4A mutations associated with familial ALS. Smith BN, et al. Neuron. 2014 Oct 22;84(2):324-31

C9ORF72, implicated in amytrophic lateral sclerosis and frontotemporal dementia, regulates endosomal trafficking. Farg MA, et al. Hum Mol Genet. 2014 Jul 1;23(13):3579-95.

A yeast functional screen predicts new candidate ALS disease genes. Couthouis J, et al. Proc Natl Acad Sci U S A. 2011 Dec 27;108(52):20881-90.

Mutations in FUS, an RNA processing protein, cause familial amyotrophic lateral sclerosis type 6. Vance C, et al. Science. 2009 Feb 27;323(5918):1208-1211.

TDP-43 mutations in familial and sporadic amyotrophic lateral sclerosis. Sreedharan J, et al. Science. 2008 Mar 21;319(5870):1668-72.

What influences patient decision-making in amyotrophic lateral sclerosis multidisciplinary care? A study of patient perspectives. Hogden A, et al. Patient Prefer Adherence. 2012;6:829-38.

Additional impact indicator information

Additional impact indicator information

(Provide information about any indicators not captured above that are relevant to the impact study, for example return on investment, jobs created, improvements in quality of life years (QALYs). Additional indicators should be quantitative in nature and include:
- name of indicator (100 characters)
- data for indicator (200 characters)
- brief description of indicator and how it is calculated (300 characters).)

Name

Public Outreach and Fundraising

Indicator Data

More than $2M donated towards MND clinic and research, from more than 17,000 individual donors.

Indicator Description

Macquarie actively seeks philanthropic support for the university’s MND-related clinical and research activities. This is co-ordinated through the University’s Office of Advancement, who manage a calendar of MND fundraising events through the year. They also manage individual donor relations.