



JOINT RESEARCH CENTRE WITH



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Director's Report

66 Medical science proved its worth in 2020 as the world grappled with the COVID-19 pandemic. CMMIT was able to contribute to this effort through research on new tests and therapeutics as well as studies on immunopathology.



Professor Steve Wilton AO
Director, Centre for Molecular
Medicine + Innovative Therapeutics &
The Perron Institute for Neurological
and Translational Science

Though less than 2 years old, the Centre for Molecular Medicine and Innovative Therapeutics - CMMIT for short - is already making its mark. With research income of \$5.2 million in 2020, 16 postdoctoral fellows and 38 PhD students, CMMIT has already exceeded the targets set at its launch in 2019.

CMMIT is unique amongst Murdoch's research centres because it is a partnership, involving researchers from both Murdoch and the Perron Institute, Western Australia's oldest medical research institute. Having a foot in both organisations is a source of considerable strength as it allows CMMIT to draw on a broader range of expertise in the area of personalised medicine. Our goal has been to create a multidisciplinary and interdisciplinary centre focused on applying the principles of personalised medicine to a range of different inherited and acquired disorders. The simple fact is every person is unique due to differences in their genetic makeup, and these differences influence the way they respond to therapies - some respond well, others poorly and still others experience adverse side-effects. The goal of personalised medicine - which some believe has the potential to transform medical practice - is to develop treatments uniquely tailored to the needs of individuals.

To create greater balance, CMMIT expanded in 2020 with the addition of two new research led by Professor Bruce Gardiner and Dr Sarah Rea. Bruce's research focuses on integrating the physical, chemical and biological processes that underlie diseases such as osteoarthritis, colorectal cancer, and acute kidney injury.

Sarah's research focuses on molecular processes in amyotrophic lateral sclerosis and other diseases.

The gamechanger for medical science in 2020 was the COVID-19 pandemic, which refocused research efforts around the world. Despite the advent of vaccines, the pandemic continues to pose a threat and this will continue for some time given mounting evidence of 'post-COVID syndrome', a group of long-term multiorgan effects seen in many patients. CMMIT showed its intrinsic adaptability by responding to the COVID-19 outbreak through a number of research initiatives, including antisense oligonucleotidebased therapeutics, new approaches to high-throughput testing for SARS-CoV-2 and studies on immune mechanisms.

If there is one feature that best defines CMMIT, it is the extent of its network of local, national and international collaborative links. Science is a global endeavour and so global links are essential to success. In 2020, not only did CMMIT researchers collaborate with over 100 institutions spread across Australia, but they co-published with colleagues from 291 institutions in 44 countries worldwide. The net result is that 62.7% of CMMIT's 2020 publications involved overseas co-authors, a figure only surpassed by three Australian universities.

CMMIT's success hinges on the continuing support of the Perron Institute and Murdoch. I would like to thank the Perron Institute and in particular its CEO, Steve Arnott and Murdoch's Pro Vice Chancellor for Health Futures Jeremy Nicholson, for being a continuing source of support and advice.

Professor Steve Wilton



Achievement & Impact in 2020



118 SCIENTIFIC PUBLICATIONS

60% in SCImago Quartile 1 journals with av. Impact Factor of 4.4 90% of publications with co-authors from other institutions

63% of publications with overseas co-authors



MAJOR GRANTS

FightMND for research on novel genetic therapies for sporadic amyotrophic lateral sclerosis (led by Anthony Akkari & Loren Flynn)

MRFF GRANT FOR A GLOBAL STUDY ON THE USE OF SIROLIMUS (LED BY MERRILEE NEEDHAM)



ANNUAL RESEARCH INCOME OF \$5.2 MILLION

28% of research funding from industry or investors \$8.4 million total income from all sources



10 RESEARCH GROUPS

16 Postdoctoral Research Scientists

38 PhD & Masters Students



Craig McIntosh & Kristin Ham

WINNERS OF THE 2020 CITY OF PERTH ASPIRE AWARD



7 NEW PATENTS ISSUED PLUS ONE PROVISIONAL (PCT)

Involvement in two spin off companies,

RAGE Biotech &

Black Swan Pharmaceuticals



COLLABORATIONS

in Western Australia, with all universities, research institutes and major hospitals

Nationally, with 105 institutions in all Australian States and Territories

INTERNATIONALLY, WITH 291 INSTITUTIONS IN 199 CITIES AND TOWNS ACROSS 44 COUNTRIES



TWO FDA APPROVED DRUGS

Two drugs developed by Perron researchers (now at CMMIT) approved for use in Duchenne muscular dystrophy

New Research Groups in 2020

Functional Genomics

The Functional Genomics group headed by Dr Sarah Rea and supported by the MNDi Foundation, Murdoch and the Perron Institute was launched in 2020. The group focuses on understanding the molecular pathogenesis of motor neurone disease, in particular amyotrophic lateral sclerosis (ALS), and frontotemporal lobar degeneration (FTLD). The group is looking specifically at genes involved in autophagy and cell signalling, including those for TANK-binding kinase 1 (TBK1) and Sequestosome 1/p62 (SQSTM1/ p62). Understanding how gene mutations cause disease helps to identify potential pathways for modulation by therapeutic strategies.



In 2020, Adriana Foster passed her PhD, becoming Dr Foster! Her research was amongst the first to show that ALS-linked mutations in SQSTM1/p62 affect cell signalling pathways in motor neurones, a finding with important implications for the field. Alistair Wood joined the group in 2020 as a Master's student. He is investigating the effects of mutations in the SQSTM1/p62 gene on stress granule dynamics and TDP-43 mobility in motor neurones. This work forms part of a Motor Neuron Disease Research Australia Innovator grant.

Nicole (Nikki) Polain also joined the group in 2020 and is investigating the effects of TBK1 mutations on cellular processes, including autophagy.



Alongside Adriana Foster and Notre Dame practicum students Eleasha Figardo and Poppy Downing, Nikki helped identify that TBK1 is a novel regulator of TDP-43 by showing that an ALS-linked TBK1 mutation fails to induce autophagy or the autophagic degradation of TDP-43. This work highlights potential mechanisms of disease for TBK1 mutations, which cause ~4% of ALS and FTLD cases. This works forms part of a Dementia Australia Research Foundation grant.

The Functional Genomics group is currently investigating new strategies for preventing p62-induced TDP-43 proteinopathy. In ALS and FTLD, TDP-43 aggregates abnormally in the cytoplasm, leading to nuclear depletion of TDP-43 and reduced TDP-43 mediated RNA metabolism. We have shown that higher p62 expression can induce TDP-43 aggregation in the cytoplasm and cause a loss of TDP-43 nuclear functions. We are exploring whether an antisense oligonucleotide-based strategy to prevent interaction between p62 and TDP-43 will stop the TDP-43 proteinopathy. We plan to test this in ALS patient-derived induced pluripotent stem cell derived motor neurones.

The Functional Genomics group is committed to collaboration. It has strong links within CMMIT with researchers such as Sue Fletcher, Anthony Akkari, Loren Flynn, and Merrilee Needham and has active collaborations with Nathan Pavlos (UWA), Bryan Ward (Department of Health), Robert Layfield (University of Nottingham), Justin Yerbury (University of Wollongong) and Brad Turner (Florey Institute).

Cell-Tissue Systems Modelling

The Cell-Tissue Systems Modelling group joined CMMIT in 2020. Led by Bruce Gardiner, the group's expertise lies in combining physical, chemical and biological processes that characterise biological systems into predictive 'systems' level computer simulations.

Generally what researchers in biomedicine face is overwhelming complexity. They are confronted with data from a wide variety of sources, from clinical trials all the way down to studies on individual molecules within specific cell types. Each of these datasets comes with its own caveats or confounding effects. Moreover, there is a tendency for biomedical researchers to stay within discipline boundaries introducing yet another layer of complexity. Biomedical problems do not easily fit into expertise silos but when we try to move beyond them our brains simply cannot cope with the volume of data available. This is where mathematical and computational modelling plays a role.

Mathematics has been described as the language of logic. Which is not to say we cannot be logical without mathematics. Instead as the number of phenomena increases, it can quickly overwhelm. Mathematics offers a framework in which to place the various bits of information into their proper context. Once a model is constructed we can then explore. Our models enable computer experiments to be performed.



Their predictive ability enables a search for treatment strategies. Our models include intra- and extracellular signalling networks, biomechanics and mechanobiology, transport processes and cell population dynamics, positioning us to tackle a wide range of problems.

To illustrate, one project with collaborators from Melbourne University, UWA and MIT focuses on articular cartilage, and how it degrades in osteoarthritis. We have been developing a series of models describing different aspects of cartilage tissue damage and repair. These models span the scale from the whole limb to sub-cellular processes. As cartilage's function is largely mechanical, it is known to respond to mechanical signals (walking is good for your knees) but it is also affected by hormones and growth factors. Over time we have developed progressively more sophisticated models of the interplay between mechanics, growth factors, cytokines, cell synthesis of cartilage components and the degradation of those components. This work has culminated in an understanding of how cartilage tissue 'works' and fails. Our work on cartilage continues. We currently hold an Australian Research Council grant focused on how cartilage achieves its fantastic slipperiness. This focuses on a layer of molecules at the cartilage surface, the brush border. In 2020 we published a paper about how long lubrication lasts after standing up. We have started dangling our legs every 30 minutes when out on a walk to decompress our cartilage!



We have a second Australian Research Council project on kidney oxygenation. This supports a talented research fellow, Dr Chang-Joon Lee. The motivating contradiction is why does a tissue that receives so much highly oxygenated blood so frequently becomes hypoxic. Normally the kidney does not extract much of the oxygen available to it. This can change, for example, during diabetes or major surgery. This project primarily involves renal physiologists at Monash and yielded two published papers in 2020. Our interests in tissue maintenance and adaption and the kidney has recently led us to explore how a fine structure called the glomerular basement membrane is maintained or changed during disease.

PROFESSOR BRUCE GARDINER WITH PHD STUDENT, AZIN AZADI AND POSTOCTORAL SCIENTIST, DR CHANG-JOON LI

Established Groups



Neurodegenerative Disorders

A major focus of the group in 2020 was analysing the role of transposable elements in Parkinson's disease (PD) and motor neurone disease (MND). The group, based at both the Perron Institute and CMMIT, used the longitudinal PPMI cohort genomics dataset, which involves whole genome sequences (WGS) from 1,300 participants, whole transcriptome data from blood for five annual visits as well as clinical and imaging data. The group now has WGS data from 212 individuals with tissue transcriptome data from brain and peripheral tissues. The group analysed the whole transcriptome profiles of PPMI PD patients and MND brain samples for regional differential expressions and for the potential impact of the transposable elements.

Grants from the Michael J Fox Foundation have allowed us to verify the tools used to identify transposable elements. A main finding from the initial PD studies is that transposable elements are linked to faster striatal neurodegeneration and faster progression of disease.

These elements are in the locus of the MAPT gene and regulate alternative splicing of several genes. New genomic variants, called SVAs, were detected in the genome of PD patients and identify patients with faster progression and whose motor performance declines quicker. This information is vital in order to plan personalised care for PD patients and shows the necessity of analysing genetic variants in PD patients before planning treatment.

The group also identified genetic loci linked to faster cognitive decline in PD. As part of a large international study of PD patients, variations in the RIMS2 gene were linked to a 4.7-fold increased risk of developing dementia. Again, as in the case of motor performance, early detection of the elevated risk for dementia in PD could provide an opportunity for interventions that postpone the onset of dementia or slow its progression. The group also showed that dementia risk is very strongly enhanced by GBA and APOE variants. This means that genetic predisposition to cognitive and motor decline in PD is linked to diverse genetic loci. This information is translatable to the clinic as it facilitates the personalised management of PD patients and improved care. The group also identified that the APOE genetic effect is caused by changed activation of TOMM40, a gene responsible for the survival of mitochondria. Taken together, these genomic studies have identified markers in PD patients that predict the progression of disease and the risk of dementia and decline in motor function. This predictive capacity will help in planning interventions to improve the quality-of-life of PD

As a new initiative, the group has embarked with colleagues on projects called TONIC-MND and TONIC-PD. These projects focus on analysing the quality-of-life of MND and PD patients in order to identify those elements that have the greatest affect. TONIC is a self-administered study that includes measures of quality-of-life not previously analysed, such as fatigue or pain in PD patients. The TONIC study tries to understand what the major impacts are and how they might be managed. The TONIC study has a potential to significantly improve the quality-of-life of patients with chronic neurological condition by offering personalised solutions for clinicians, carers and family members.

In addition to the work on neurodegenerative diseases, the group is working in developing antisense oligonucleotide-based drugs for psoriasis and osteosarcoma.



Demyelinating Disorders

Headed by Clinical Professor Allan Kermode, the group continued to receive generous support from MSWA as a part of its state-wide support for research and clinical development for multiple sclerosis. The group was established many years ago at the Perron Institute and has a growing presence at Murdoch University. Of particular note, Allan Kermode has recently been appointed Deputy Director of the Professional Services Review, and Chair of its Neurology Panel, an independent Commonwealth Statutory Agency directly responsible to the Minister for Health for the regulation and integrity of Australia's MBS and PBS system.

The funding from MSWA enabled continuation of research on biomarkers (e.g. neurofilament light chain) and molecular and radiological aspects of MS conducted by Marzena Fabis-Pedrini. Clinical research also continued on epidemiology, genetic determinants of disease, immunogenetic and phenotypic relationships, prognosis, biomarkers and imaging studies, as well as curation of clinical and laboratory data. Dr Fabis-Pedrini has analysed changes in NfL levels in a large longitudinal cohort and has begun quantitative MRI analysis. A case control comparison study with MS patients in Southern China is also underway and efforts are being made for the group to contribute a greater number of cases to the MSBase collaboration, with Claire Tucak coordinating data entry into MSBase.

Immune dysregulation in early MS, including the immunomodulatory effects of ultraviolet light, is being studied by Stephanie Trend. Dr Trend is examining changes in immune cell types at the onset of MS, and identifying changes in their immune activation and regulation. She is particularly interested in changes in B-cells, and has been awarded a MSRA grant to further these studies. The primary focus of sample collection is drug naïve recently diagnosed individuals with MS or CIS.

An exciting new collaboration has commenced with Prof Georges Verjans, Erasmus Medical Centre (Netherlands), on immediate post-mortem MS brain tissue. This uses the technique of single-cell sequencing and has identified relevant CD4+ and CD8+ T-cell TCRs in MS lesions and also in apparently normal white matter. Transcriptomics have been performed.

This work is being undertaken by Dr Xiaonan Zhong, a neurologist from Sun-yat Sen University, in China who is enrolled in a PhD with Professor Kermode. Her goal is to find the target antigens using both unbiased (T-scan) and specific strategies. The group will be assisted in this work by David Koelle (Washington University), and Simon Mallal (Vanderbilt University). The TCR/antigen-HLA complexes will be identified, and we will confirm enrichment in our large MS cohort. The results will feed into the clinical and immunogenetic datasets being prepared by Dr Fabis-Pedrini, and the cellular immunological studies of Dr Trend. Finally, we will assess the influence of this complex on MS disease severity, clinical features and treatment response.

The group published 26 scientific articles in 2020 in high-impact factor journals, including Lancet Neurology, Scientific Reports, Frontiers Neurology, Multiple Sclerosis and Neurology. Despite the ongoing COVID-19 pandemic, the group also gave virtual presentations at several international and national scientific meetings.

A new collaboration with the University of Newcastle has been established that will result in analysis of HLA A, B, and C data in our MS cohort. The Group has an active network of collaborations with the Chinese University of Hong Kong (Alexander Lau), Sun Yat-sen University (Wei Qiu), Switzerland (Jens Kuhle), UCSF (Jorge Oksenberg), Erasmus Medical Centre Netherlands (Georges Verjans), Vanderbilt Medical Centre (Simon Mallal) and Sweden (Bjorn Frendeus) as well as ANZGene, IMSGC and Regeneron. Multiple publications have resulted as a consequence of these productive collaborations.



Motor Neurone Disease

The Motor Neurone Disease (MND) group continues to develop personalised genomic medicines for MND patients and was greatly aided in this by two competitive grants awarded in 2020 for research on antisense therapeutic targeting the SOD1 gene. The grants were from the Motor Neurone Disease Research Institute Australia (MND-RIA) to explore the broader applicability of the SOD1 therapy in sporadic MND, and from FightMND to conduct preclinical toxicology/safety studies to progress the SOD1 therapeutic through to the point of US Food and Drug Administration (FDA) engagement, towards Phase 1.

The work of Dr Loren Flynn in advancing antisense therapies (SOD1 and others) has been a vital element towards achieving the group's goal of making a difference for patients with MND. The collaboration between A/Prof Turner (Florey Institute), Dr Flynn and Prof Akkari continues to strengthen. The SOD1 therapeutic is now being tested in animal models of MND and has been shown to reduce levels of the toxic form of the SOD1 protein. This work commenced recently and the therapeutic dose is currently being optimised to allow future studies of safety.

In 2020, the MND group published several scientific papers on the importance of a class of genetic markers known as structural variations (SVs). These raised important questions about how to identify new genetic markers for sporadic MND in areas of the human genome that have been previously under investigated. Later in 2020, the group published two papers showing that SVs are associated with both familial and sporadic MND. The work on SVs has continued to grow and several promising genetic markers that affect the phenotype of sporadic MND have been investigated. This has led to significant collaborations developing with Professor Ammar Al-Chalabi and his team at Kings College London, and Professor Don Cleveland at the Ludwig Institute in San Diego.

The MND group continued to strengthen its links with industry, progressing collaborations with two start-up companies, GenieUS (Australia) and Black Swan Pharmaceuticals (USA). GenieUS is developing therapeutics and diagnostics for sporadic MND. Through the collaboration with GenieUS, the MND group plans to develop novel microRNA therapies for MND patients. Black Swan Pharmaceuticals is an antisense therapeutics company, established to translate the therapies developed by the MND team into Phase 1 for sporadic MND and other neurodegenerative diseases, such as Parkinson's disease.

The MND group has a strong commitment to collaboration. Aside from its grants with the Florey Institute and Black Swan Pharmaceuticals, collaborations continue with the Division of Neurology at Duke University, Northwestern University, Griffith University (Professor Alan Mackay Sim), and Kings College London (Professor Ammar Al-Chalabi). These collaborations will form the basis for future grant applications and provide a vehicle for placement of our team members. Of significant note, Professor Akkari was invited to join the National MND Summit Committee as the WA representative. The purpose of this newly formed organisation is to bring together MND researchers from around the country to develop a national strategic plan for MND research

In 2020, Professor Akkari and Dr Flynn attended the Neurodegenerative Disease Drug Development Conference (Boston) and presented on their research on novel splice-switching molecular therapeutics and genetic markers for amyotrophic lateral sclerosis (ALS). In December 2020, the MND group were a major presence at the on-line 31st International Symposium on ALS/MND, where four group members both presented posters and gave 'live' video presentations. The group is particularly proud of the fact that, at the Perron institute's 3-Minute Thesis Competition, the first, second and third places all went to PhD students from the MND group.

On a final note, the group wishes to acknowledge the ongoing support of the Giumelli Family Foundation for the MND cell model laboratory and the Racing for MNDi Foundation.

Molecular Therapy

The Molecular Therapy group had another productive year and, like most at CMMIT learnt to be more agile and responsive to the changing world situation. COVID-19 lockdowns saw many of our team working from home sometimes for weeks at a time. However, this did not hinder our successes as we used the time to plan, write papers, apply for grant funding, and of course further our research in developing novel therapeutic strategies to modify gene expression.

Never one to miss out on opportunity, we extended our collaboration with colleagues at Monash University, starting with converting the pro-inflammatory membrane bound form of RAGE (receptor for advanced glycation end-products) to the potent anti-inflammatory soluble form. We extending this strategy to the surface bound ACE2 (angiotensin converting enzyme 2, which acts as the entry point for coronavirus infections) by promoting expression of a soluble ACE2 isoform that can act as a virus decoy. The preliminary work was sufficient in securing an MRFF Antiviral Development for COVID-19 Grant in collaboration with the Monash group.

Our group furthered cemented its relationship with Sarepta Therapeutics through a Sponsored Collaborative Research Agreement for another three years. Our research with Sarepta on neuromuscular and neurological diseases remains a major driving force shaping current and future research projects.

The group congratulates Professor Sue Fletcher on her appointment to the role of Chief Scientific Officer at PYC Therapeutics. Due to the demands of her new position, Sue decided to step back from her role as a Principal Investigator as part of the Murdoch-Sarepta partnership. However, we are not losing her completely as she will retain a partial appointment at Murdoch as a Senior Principal Fellow.

Although we only took on one new staff member, 2020 was nonetheless yet another year of growth in terms of leadership and the expanding roles of many of the group's existing staff and students.

Dr May Aung-Htut was promoted to Senior Research Fellow and took on additional leadership and mentoring roles within the group. Her paper "Splice modulating antisense oligonucleotides restore some acid-alpha glucosidase activity in cells derived from patients with late-onset Pompe disease" further confirms the potential of antisense therapy as a treatment option for this progressive disease.

Kristin Ham, who has been a vital part of our team since 2012, has taken on the added challenge of doing a PhD. We look forward to the results of her research using antisense oligonucleotide modification of COL7A1 transcript expression as a therapy for the devastating disorder, recessive dystrophica epidermolysis bullosa. Craig McIntosh officially graduated and took on a postdoctoral scientist role in the group after completing his PhD on antisense oligonucleotide-mediated therapeutic strategies for neurodegenerative repeat expansion disease. Kristin and Craig's hard work was recognised by their award as joint winners of the City of Perth Aspire Award. The Aspire Awards are designed to support Western Australian researchers, academics, and professionals to attend an international business event as a means of professional development. This is truly a great opportunity for these young researchers and the group was thrilled that Kristin and Craig received this recognition. Dunhui (Oliver) Li also completed his PhD and Jessica Cale is following closely behind. Several other students, Janya Grainok, Sasiwoman Utama and Bal Poudel, are due to submit in 2021. Dunhui's paper "A splice intervention therapy for autosomal recessive juvenile Parkinson's disease arising from parkin mutations", the first published by the group on Parkinson's disease, successfully showed functional rescue parkin protein, suggesting that a splice switching antisense strategy, originally developed for Duchenne muscular dystrophy, may be another candidate for clinical trials in the near future. Even though the group moved from the QEII campus to Murdoch seven years ago, it still sees itself as an integral part of the Perron Institute and views the creation of CMMIT as a way of expanding its network of collaborations.



CENTRE FOR MOLECULAR MEDICINE + INNOVATIVE THERAPEUTICS >> ANNUAL REPORT 2020

Myositis

Despite the changing and challenging landscape driven by COVID, 2020 has been an important year for expansion of the Myositis group. We have continued to establish a strong foundation for our research programme at both CMMIT and the Perron Institute, strengthening our patient partnerships and collaborations and investing in personnel to facilitate growth. It has also been an exciting year of grant success, with the team securing a \$1.8M NHMRC/MRFF grant to lead an international, multi-centre Phase 3 clinical trial of Sirolimus in Inclusion Body Myositis (IBM).

In 2020, our team expanded both the laboratory and clinical arms of the programme, with Physiotherapist Ian Cooper, Research Coordinator Monica Lam and Software Developer Matthew Needham joining the clinical team, and Anu Sooda joining the laboratory team. Together with the important contribution of our students, the addition of these team members allowed us to expand our biobank resource significantly. The biobank now includes over 300 consented myositis patients and healthy controls, providing a valuable resource for our group and also facilitating national and international collaboration. Many of our myositis patients continue to provide repeat samples for our biobank during their clinic visits, enabling longitudinal analysis of immune response. 2020 has also seen a growth in our 'myobank', with patients undergoing diagnostic muscle biopsies kindly donating samples for our research.

The laboratory programme, led by Dr Jerome Coudert, supported four students in 2020 undertaking PhD, Masters and Honours degrees. Murdoch University students, Nataliya Slater and Emily McLeish both secured PhD scholarships and Murdoch University Honours student Andrew Wallhead started his research project, while UWA student Alice DiVincenzo completed her Masters project obtaining a high distinction. Anu Sooda, who started with the group in 2020, brought in a methodology toolset particularly well-fitted for the research programme.

Alongside the Myositis Discovery Programme projects, Jerome Coudert started his involvement in three collaborative projects with other CMMIT researchers.

During the first COVID lock-down in March-April 2020, students analysed their data and worked on their theses, thereby optimising the time away from the lab, while Dr Coudert applied his expertise in Immunology to submit multiple grant applications for SARS-Cov2 research and initiated new collaborative projects with Jeremy Nicholson's and Elaine Holmes' groups at ANPC. This work has provided evidence of immune-metabolic signatures in individuals severely affected by COVID-19 and so far has resulted in two publications in Journal of Proteome Research and one publication in the top-tier Immunology journal, Immunity.

Within our clinical arm, we now have a well-established model for integrating research and clinical care for patients attending clinics at Murdoch and the Perron Institute. Patients benefit from clinic visits that include in-depth assessment of strength and function by our Physio, Ian Cooper, as well as social and chronic disease support from our Nurse, Kelly Beer, and Research Coordinator, Monica Lam. Alongside this, patients (and often their partners/family members) contribute data and samples during their visits. Patients have been incredibly supportive of this model of care and we have plans to measure the positive impact of these clinics and share practice.

We continue working to optimise tracking of the natural history of disease and alongside outcome measures collected in clinic, are working to integrate our novel database, the Myositis Registry into clinical practice. We now have over 70 patients consented to the Registry and are currently in a final software development phase. The plans for roll-out of the Registry across Australia are underway, with national ethical approval (NMA) granted.

Bi-directional, patient-centered research continues to be a focus and in 2020 we continued to work closely with the Myositis Association of Australia to close the divide between patients (consumers) and researchers, including facilitating an Australia-wide Q&A session with leading myositis clinicians and patients during Myositis Awareness Month in May. We also managed (amongst COVID restrictions!) to host our much-anticipated annual Research Update day in September 2020. This day was once again a great success, with over 80 attendees.





Clinical Exercise and Cognition

Unlike CMMIT's other research groups, the Clinical Exercise and Cognition group (CEC) brings together a diverse group of research academics from the disciplines of Psychology and Exercise Science at Murdoch University. These researchers share a common interest in implementing lifestyle interventions and applying non-invasive techniques to improve the quality of life of individuals living with a range of chronic conditions. The group's members are primarily research and teaching academics, which allows many of the research outcomes to be directly transferred into the classroom environment. This not only enriches the learning environment for students but also encourages graduating students - primarily ones destined to become clinicians in the fields of Exercise Physiology or Psychology - to engage in research in this area, and ultimately build research and clinical capacity in this area.

The close integration of the CEC group's research and its teaching activities is unique within CMMIT. The group's interests are broad and encompass both basic research and research using human subjects. Amongst the disorders under investigation are obesity and obesity-related morbidities (including type 2 diabetes and insulin resistance), multiple sclerosis, inflammatory myopathies, brain injury (including stroke), age-related decline in cognitive and motor function, Alzheimer's and Parkinson's disease, sports injuries, cardiovascular disease, and musculoskeletal disease. The group includes exercise physiologists, neuro-physiologists, physiotherapists, and exercise and sports scientists.

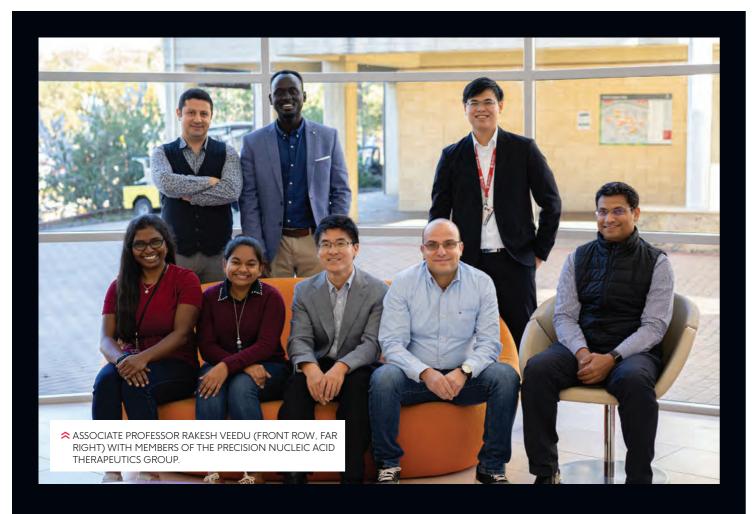
In 2020, the group published 20 scientific papers, equivalent to 17% of CMMIT's total publications. Of these, approximately 47% were in SCImago Q1 journals with over 13% in journals ranked in the top 10% cited worlwide.

These publications cover topics as diverse as visual-perceptual training and visual anticipation, the relationship between physical activity, cognitive health and neurodegenerative change in the brain, cardiorespiratory fitness and memory, the benefits of physical activity in multiple sclerosis, exercise training and muscle size, strength, and function in older adults and non-invasive brain stimulation. The scope of the group's research illustrates not only the diversity of the CEC group but also its potential over time to feed into CMMIT's clinical research agenda.

Two of the group's PhD students completed in 2020 and members of the group supervised 12 Masters and PhD students and 10 honours students. The involvement of the CEC group in CMMIT has been a positive catalyst for research in the group with several researchers focusing some of their research towards the neuroscience area, while those previously engaged in the neurosciences, were able to achieve research translation through the improved access to clinicians and patients that CMMIT provides. The shift in research focus was also evidenced by the topics of research pursued by incoming Master of Exercise Science (Research) students, where two of the three projects involved participants with Parkinson's disease, multiple sclerosis or myositis. These students will develop skills, interests and knowledge in exercise prescription for individuals with neurological conditions, thereby increasing clinical and research capacity within the area within Western Australia. We anticipate this trend to continue throughout 2021.

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Precision Nucleic Acid Therapeutics

Although largely funded by the Perron Institute, the Precision Nucleic Acid Therapeutics (PNAT) group is primarily based at Murdoch University where it forms part of a larger research grouping in the area of nucleic acid therapeutics. The group's research focuses on developing novel nucleic acid-based therapies and diagnostic devices to tackle a range of rare and acquired diseases. For this purpose, the group's research activities can be categorised into three main areas: 1. oligonucleotide synthesis chemistries; 2. oligonucleotide drug and diagnostic development; and 3. oligonucleotide drug delivery. Most of the group's research is commercially focused. In 2020, the group comprised of 9, including 4 postdoctoral fellows, 3 PhD students and 2 Honours students.

The PNAT group made significant progress in 2020. Firstly, Madhuri Chakravarthy's PhD was conferred in August. Similarly, Suxiang Chen completed and submitted his PhD thesis. A new PhD student, Arpitha Chikkanna joined the group in January to undertake a project aimed at developing antisense therapy for cancers. Also, Isaac Ronyo began his Occupational Traineeship towards the end of 2020 prior to commencing BSc Honours.

The PNAT group published or co-authored 9 articles in internationally recognised journals. The group received funding from a variety of sources in the form of both grants and industry funds. It received funding from the Western Australian Department of Health through its COVID-19 program to develop a rapid diagnostic kit for SARS-CoV-2 using lateral flow paper test strips and Rakesh Veedu was awarded a Western Australian Department of Health Merit Award for research on next generation morpholino oligonucleotides for therapeutic alternative splicing. The group's COVID-19 research was featured in the media in an article in the West Australian and on ABC TV's 7:30 Report.

Through a developing research collaboration with Glen Reid and Sarah Diermeier from the University of Otago in New Zealand, the PNAT group was successful in obtaining funding from the Ministry of Business Innovation and Employment aimed at building new capabilities in the field of functional nucleic acids to fight disease. Research continues on a novel trivalent GalNAc phosphoramidite for liver-targeted delivery of antisense oligonucleotides. In addition, the group continues to receive generous support for commercial research from Dr George Bautovich and the Caruthers Family Foundation.

Blood Disorders

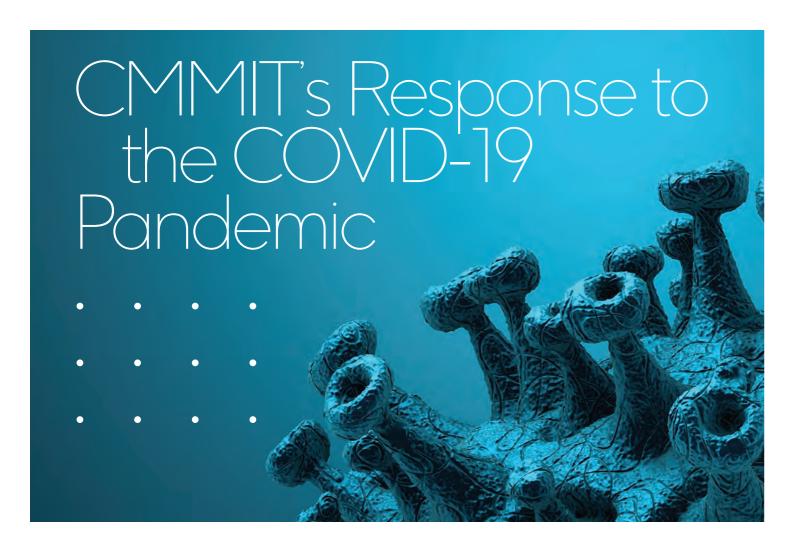
It has been an extraordinary past 12 months and like many facets of life around the world, the once in a life-time COVID-19 pandemic impacted heavily on the Blood Disorder group's research activities. During the first half of 2020, research activities were paused for the majority of wet-laboratory research as well as clinical trials. Instead, the focus was shifted to crystalising research results into published manuscripts, as well as grant applications and the development of new ideas.

One positive outcome was the publication of the group's research on microRNA-365a-3p and its regulation of pro-coagulation protein Tissue Factor in response to different levels of oestrogen. MicroRNAs are small but powerful gene regulators in our body. Their effects can be rapid, and some out-of-control microRNAs can be highly detrimental, e.g. cause cancer or increase the likelihood of developing a blood clot. In our paper, we showed an association between high physiological oestrogen levels during pregnancy and hormone-based contraceptive use, which led us to comment on the further research needed to corroborate this association. The project underlying the paper is part of Jiayin Tian's PhD dissertation and the whole group was thrilled for Ms Tian that her research should have resulted in such important findings. After several months of review, the paper was published in the prestigious journal Thrombosis and Haemostasis, one of the highest ranked journals in the field of haematology. Following her publication success, Ms Tian submitted her PhD dissertation for examination. We await the outcome of the PhD thesis examination process. Other publications resulting from collaboration with national and international groups include a study examining the efficacy and safety of dual therapy in the treatment of a type of cancer in the lymph nodes called non-Hodgkin lymphoma (CHRONOS-3), which was published in the prestigious journal, Lancet Oncology. The study was the first to show broad and superior efficacy of copanlisib plus rituximab in patients with relapsed indolent non-Hodakin lymphoma. Other important publications include a SWAN study examining VTE patient satisfaction after conversion from the traditional blood thinner, warfarin, to the newer and easier to manage direct oral anticoagulants.

In addition to clinical trial and laboratory studies, the Blood Disorder group was active in the quality assurance network in laboratory medicine. A recent publication, in collaboration with national and international collaborators, highlighted the importance of laboratory blood measurements in studies on new generation of anticoagulants called DOACs. The group's contribution to the international community in the area of thrombosis and haemostasis has thus translated into better patient care. Our research in thrombosis and haemostasis has fed into several haematology working groups on ADAMTS13 measurements, focusing on a protein, von Willebrand Factor, important in guiding treatment in a class of rare diseases. von Willebrand Factor is important for clotting during injury, as well as vaccine-induced prothrombotic immune thrombocytopenia (VIPIT) - also referred to as Thrombosis with Thrombocytopenia Syndrome (TTS) - as seen in selected, albeit rare, recipients of the COVID-19 Astra-Zeneca vaccine. The group continues to foster international collaborative research through the IMATAS study, as well as on other key thrombosis and haemostasis related topics.

The enforced time-out in wet-laboratory work has reinvigorated the group's research. Whilst working from home, we conducted brain-storming Zoom sessions to bounce ideas on the future direction of our research. We believe our partnerships and collaborations across the Murdoch University research community as well as externally will be the capstone for success in 2021.





Most scientific research is steady-as-you-go, following an often preset schedule of experiments. Occasionally, however, you need to drop everything and turn your attention to a pressing challenge. Such was the case in 2020 when the COVID-19 pandemic struck.

The pandemic - a once in a century event that to date has led to nearly 4 million deaths worldwide – has required researchers across multiple fields to redirect their skills to finding solutions to this pressing public health issue. CMMIT researchers proved their adaptability by being part of this effort, as shown in the examples below.



A rapid paper test for SARS-CoV-2

In partnership with the Fiona Stanley Hospital and CERI, CMMIT's Precision Nucleic Acid Therapeutics group led by Rakesh Veedu has recently developed a novel lateral flow paper test for the presence of SARS-CoV-2 in swabs, which is both highly accurate and yields results in minutes.

This test uses a novel aptamer against the recombinant SARS-CoV-2 spike protein where the presence of SARS-CoV-2 is detected using an immobilised red-coloured spike protein aptamernanoparticle-conjugate plus secondary aptamer. The test is currently being evaluated for sensitivity by researchers in Singapore and is the subject of a provisional patent application. This research was supported by the Government of Western Australia's COVID-19 Special Initiative Scheme.



The immune-metabolite profile of SARS-CoV-2

In a collaboration study with the ANPC, CMMIT's Myositis group led by Merrilee Needham and Jerome Coudert has investigated the profile of inflammatory cytokines and chemokines in the blood of SARS-CoV-2 patients. SARS-CoV2-positive patients were compared with SARS-CoV2-neagtive patients with influenza-like symptoms and healthy controls.

A panel of 34 inflammatory cytokines and chemokines were shown to be significantly different in SARS-CoV2 patients. Moreover, the cytokine profile correlated with the altered lipoprotein profiles in SARS-CoV2 patients. The data strongly suggest a major role of cytokines and chemokines as well as lipoproteins in the overall immune response to the disease. This research was supported by the Government of Western Australia's COVID-19 Special Initiative

A fast and high-throughput test for SARS-CoV-2

CMMIT's Neurodegenerative Disorders group led by Sulev Kõks is currently developing a high-throughput testing technology for COVID-19 detection in patient samples that can be used in population-wide screening to control and prevent the spread of the SARS-CoV2 infection.

This test combines a fast molecular technology, easy sample collection and robotics into a consolidated platform. The test uses saliva instead of nasopharyngeal swabs and gives an answer within 30 minutes of saliva collection. The testing system has the capacity to analyse 5,000 samples per hour and is thus suitable to tackle major outbreaks of COVID-19 or be deployed in crowded settings such as airports, stadiums, borders and seaports to provide safe and rapid community monitoring. This research was supported by the Government of Western Australia's COVID-19 Special Initiative Scheme.



Inhaled oligonucleotides to generate a decoy receptor for SARS **Coronarvirus-2**

In collaboration with Monash University, CMMIT's Molecular Therapy group led by Steve Wilton has repurposed the same 'gene patch' technology used in the treatment of Duchenne muscular dystrophy to modulate expression of two genes of immense significance in human health.

RAGE is a cell-surface protein that, when activated results in a strong pro-inflammatory response. Specific 'gene patches' have been developed that change the RAGE expressed to a shorter soluble form that is potently anti-inflammatory. A spin-off company, RAGE Biotech has launched to commercialise this technology. A second application of this technology has been its use to convert the membrane-bound form of the ACE2 receptor-the receptor mediating coronavirus entry into cells – into a shorter soluble isoform that acts as a COVID-19 decoy. Preliminary in vitro experiments have shown that treated cells were highly protected against COVID infection compared to untreated cells. The research is funded through the MRFF as part of the Australian Government's \$8 billion Coronavirus National Health Plan.





The ultimate goal of all medical research is to improve outcomes for patients. Recognising this, there is increasing awareness of the need for consumer and community involvement in all phases of the research process.

With this in mind, CMMIT with the assistance of the Western Australia Health Translation Network Community Involvement Program (CCI), formed a Consumer Advisory Group in 2020, a first for Murdoch. This group is comprised of individuals with lived experience of some of the debilitating medical conditions under investigation by CMMIT researchers.

Involving individuals with direct experience of medical conditions that are of importance to CMMIT is crucial, as it enables consumers to positively influence the way CMMIT conducts its research. We believe that we need to bridge the gap between researchers and the broader community so that our research can focus more sharply on outcomes that directly benefit patients.

With assistance from our Consumer Advisory Group, we plan to review our research programs and, in the process, provide research updates aimed at the broader community so that there is greater community awareness of the research we undertake. Our goal is to facilitate public debate and to advocate for change, particularly regarding the translation of research into outcomes such as new diagnostics and therapeutics and improvements in clinical practice and policy. CMMIT views the Consumer Advisory Group as a way of increasing public understanding of the importance of health and medical research to Australia's healthcare system, economy and community.

Involving the broader community in shaping our research is crucial if CMMIT is to succeed in its mission of translating findings from the laboratory into outcomes that directly benefit patients.

- Professor Steve Wilton, Director of CMMIT



Future Science Conference

Hosted by Murdoch University in conjunction with Science Teachers' Association of WA, Future Science 2020 Conference showcased Murdoch's enthusiastic experts from science, technology, engineering and mathematics to Science Teachers, Student Teachers and Laboratory Technicians. CMMIT researcher's helped them explore cutting edge science and activities they could take back to the classroom to engage the next generation of scientists.

)> CMMIT DIRECTOR, PROFESSOR STEVE WILTON WAS THE KEYNOTE SPEAKER FOR THE DAY VIA ZOOM.



Transformers Virtual STEM careers forum 3 - 6 August

Dr May Aung-Htut and Dr Craig McIntosh represented CMMIT at Transformers - a virtual STEM Careers Forum for Year 9 to 12 secondary school students.

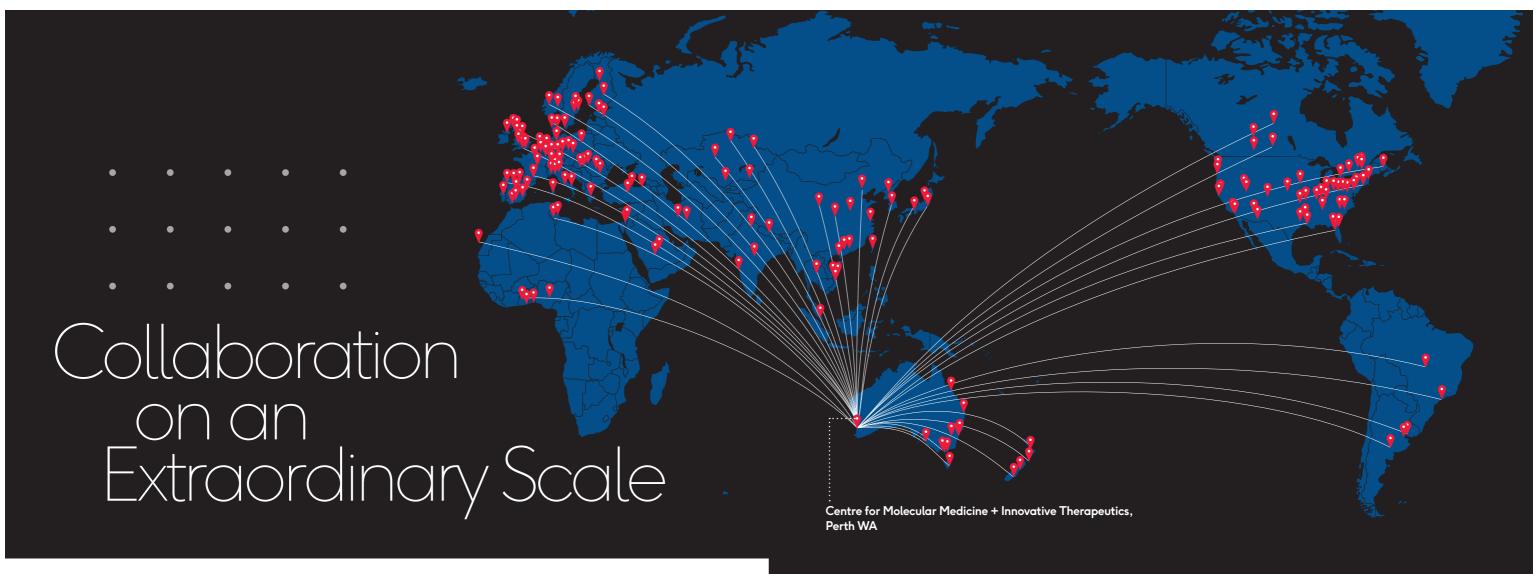
The forum provides opportunities for students to hear from, and talk with industry experts and explore post-school options in STEM Careers, inspiring the next generation of scientists

Murdoch's 2020 Science Experience

CMMIT delivered a series of workshop's to high school students, giving a taste of a career in Science, Technology, Engineering and Mathematics (STEM).



STUDENTS TAKING PART IN CMMIT'S WORKSHOP AS PART OF MURDOCH'S 2020 SCIENCE EXPERIENCE



Success in scientific research depends on collaboration with researchers and institutions worldwide. One of CMMIT's special strengths is the depth and breadth of its collaborative links.

First and foremost, CMMIT itself is a collaborative venture between the Perron Institute and Murdoch University. Secondly, collaboration with industry increasingly underpins much of CMMIT's research, particularly with the US-based biopharmaceutical company, Sarepta Therapeutics.

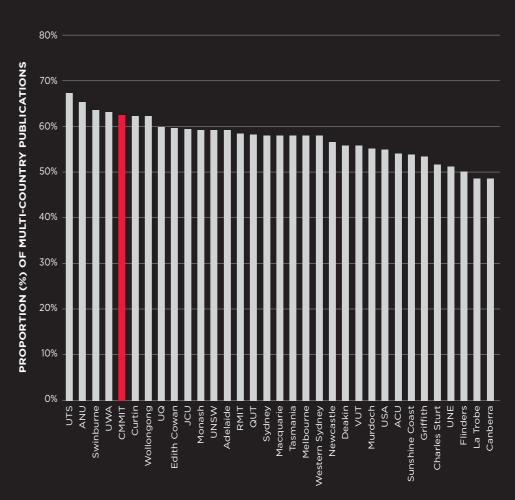
On a broader scale, CMMIT has an exceptionally strong record of research collaboration. The map above shows the global spread of CMMIT's links based on co-authored publications, joint PhD students and membership of international research consortia. In summary:

- >> LOCALLY CMMIT collaborated with all of Western Australia's universities, medical research institutes and major hospitals.
- >> NATIONALLY CMMIT collaborated with 105 institutions from all Australian States and Territories.
- >> INTERNATIONALLY CMMIT collaborated with 291 institutions in 199 cities and towns across 44 countries worldwide.

To put CMMIT's collaborative links in context, 89.8% of CMMIT's 2020 publications were co-authored with colleagues from other institutions and 62.7% involved co-authors from at least one other country. Only three Australian universities (CWTS Leiden Ranking) exceeded CMMIT in 2020 in terms of the collaborative publications and only four had a higher proportion of multi-country publications.

The chart, right, shows CMMIT's performance in terms of multicountry publications compared to Australia's universities.

- 66 Science is a global endeavour. Ideas transcend national borders and in an increasingly interconnected world, international collaboration is absolutely essential if Australian science is to flourish
 - Professor Steve Wilton, Director of CMMIT





Demyelinating Diseases

Clinical Professor Allan Kermode Group Leader

Clinical Professor William Carroll Consultant Neurologist

Dr Jason BurtonConsultant Neurologist

Dr Marzena Fabis-Pedrini

Postdoctoral Fellow

Dr Stephanie TrendPostdoctoral Fellow

Katherine Roberts

Research Assistant

Dr Xiaonan Zhong PhD Student

Claire Tucak

Research Nurse

Shaila Jereedy

Research Coordinator

Administration

Professor Steve WiltonDirector

Professor Norman Palmer

Deputy Director

Jodie Williamson Centre Manager

20

Brianna O'DonnellAdministrative Officer

Motor Neurone Disease

Professor Patrick Anthony Akkari Group Leader

Dr Loren Flynn

Postdoctoral Scientist

Dr lanthe Pitout

Postdoctoral Scientist

Nik Gavriel Bree Bell

Research Assistants

Rita Mejzini Julia Pytte

Frances Theunissen Leanne Jiang

PhD Students

Kai Plunkett Honours Student

Cell-Tissue Systems Modelling

Professor Bruce GardinerGroup Leader

Dr Chang-Joon LeePostdoctoral Scientist

Azin Azadi PhD Student

Functional Genomics

Dr Sarah Rea Group Leader

Dr Adriana Foster

Research Assistant
Nikki Polain
Research Assistant

Alistair Wood Masters Student

Molecular Therapy

Professor Steve WiltonGroup Leader

Professor Sue Fletcher

Principal Senior Research Fellow

Dr May Aung-Htut Senior Research Fellow

Dr Craig McIntosh

Postdoctoral Scientist

Abbie Adams

Senior Research Officer

Russell Johnsen Senior Research Officer

Kane Greer Research Officer

Kristin Ham

Research Officer **Dunhui (Oliver) Li**Research Associate

Alanis Lima Research Assistant

Maria Van Loenhout Research Assistant

Di Huang Kelly Martinovich Janya Grainok Khine Zaw Jessica Cale Niall Keegan

Bal Hari Poudel Sasiwimon (Fern) Utama Sarah Leishman

Leon Larcher PhD Students

Neurodegenerative Diseases

Professor Sulev Kõks

Group Leader

Dr Abi Pfaff

Postdoctoral Scientist

Dr Vidya Saraswathy Krishnan Postdoctoral Scientist

Lewis Singleton Research Assistant

Emel Rothzerg

Emel Rothzerg
PhD Student

Myositis

Professor Merrilee Needham Group Leader

Professor Frank Mastaglia

Senior Advisor

Dr Jerome CoudertPostdoctoral Reserach Fellow

Kelly Beer

Clinical Research Manager

Dr Anuradha Sooda Research Assistant

Dr Shereen Paramalingam Nataliya Slater Emily McLeish PhD Students

Alice Di Vincenzo

Masters Student

Andrew Wallhead
Honours Student

Precision Nucleic Acid Therapeutics

Associate Professor Rakesh Veedu

Group Leader

Dr Bao LePostdoctoral Scientist

Dr Tamer KosbarPostdoctoral Scientist

D-T--W---

Dr Tao WangPostdoctoral Scientist

Associate Professor Lanmei Chen Visiting Fellow

Nabayet Sbuh

Research Assistant

Akila Balachandran Suxiang (John) Chen Prithi Raguraman Arpitha Chikkanna

PhD Students

Isaac Ronyo Occupational Trainee

Blood Disorders

Prof Ross Baker

Group Leader

Dr Jim Tiao

Scientific Lead, Laboratory Manager

Dr Omar Elaskalani Postdoctoral Scientist

Grace Gilmore

Senior Research Assistant

Pat Metharom
Platelet Group Lead

Clinical Exercise Cognition

Academic Staff, Psychology & Exercise Science

Dr Yvonne Learmonth

Group Leader/Senior Lecturer

Associate Professor Tim Fairchild

Associate Professor, Sport & Exercise Science

Dr Alasdair Dempsey

Interim Dean, Learning and Teaching

Dr Ann-Mare Vallence

Senior Lecturer

Dr Hakuei Fujiyama

Senior Lecturer

Associate Professor Jeremiah Peiffer

Dean, Graduate Research School

Dr Sean Muller

Associate Professor, Sport & Exercise Science

Dr Shaun Teo

Postdoctoral Scientist

Steve Smith Nathan Smith

Kym Wansbrough

Brittany Rurak Lucy Schouten

Jane Tan

Khaya Morris-Binelli

Auretta Kumar PhD students

Jiayin Tian PhD Student

Elijah Callis

Madison Hager Honours Students

ours students



CMMIT exceeded expectations in terms of grants and industry funds in 2020, bringing in \$12.1 million in total funds. These generated research income of \$5.2 million—an increase of 126% compared to 2019. 28% of CMMIT's research funding came from industry and investors, reflecting CMMIT's strong commercialisation focus.

In addition to the above funding, Professor Sulev Kôks continued to be involved in European Commission ERA grants worth over \$21 million.

Some headline successes in research funding in 2020 were:

- >> Continued support from the US pharmaceutical company, Sarepta Therapeutics for a research program involving the Molecular Therapy group focusing on the development of novel antisense oligonucleotide-based therapeutics.
- >>> Breakthrough funding of research on genetic therapies for motor neurone disease with a \$1 million grant from FightMND to the Motor Neurone Disease group.

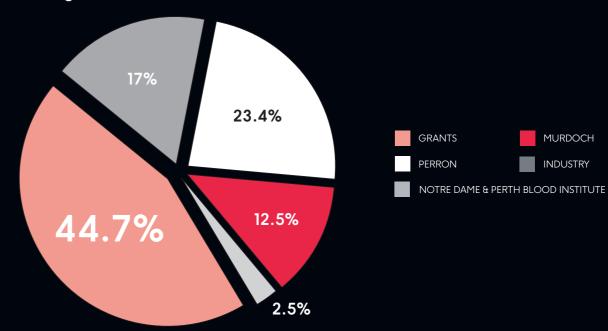
- >>> Despite WA receiving only 4.6% of national Medical Research Future Fund funding, two CMMIT groups obtained substantial MRFF grants in 2020 – the Myositis group received a \$1.8 million grant for a global study investigating the use of the re-purposed drug, Sirolimus in the treatment of Inclusion Body Myositis, and Steve Wilton is co-investigator on a grant on antisense therapies targeting the ACE2 receptor as a treatment for COVID-19.
- >>> Funding from Government of Western Australia's COVID-19 Special Initiative Scheme for a series of projects.
- >> Funding from the Michael J Fox Foundation for a study by Sulev Kôks on genetic factors that predict disease risk and progression in Parkinson's disease.

Core Funding

Aside from research grants and industry funds, CMMIT's core funding comes from its joint venture partners, Murdoch University and the Perron Institute as well as Notre Dame University and the Perth Blood Institute, which provide targeted support for research by the Myositis and Blood Disorders groups, respectively. **66** Core funding from the Perron Institute and Murdoch University provides CMMIT with a foundation which enables it to undertake cutting-edge research supported by funds from competitive grants and industry

- Professor Steve Wilton, Director of CMMIT

Funding Chart





In 2020, the share of funding from the various sources was:

\$3.764 M Competitive grants Industry funds \$1.431 M Perron Institute \$1.970 M

\$1.050 M (+ academic salaries) **Murdoch University**

Notre Dame/Perth Blood Institute \$0.21 M **TOTAL** \$8.423 M



CMMIT's output of scientific publications - the 'currency' of science - continues to increase with 118 publications in 2020, an increase of 10.3% compared to 2019. In terms of journal quality, 60% of publications were in SCImago Quartile 1 (Q1) journals and 25.4% in Q2 journals. The average impact factor (IF) of journals was 4.404.

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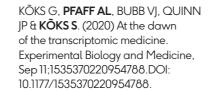
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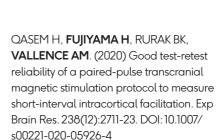
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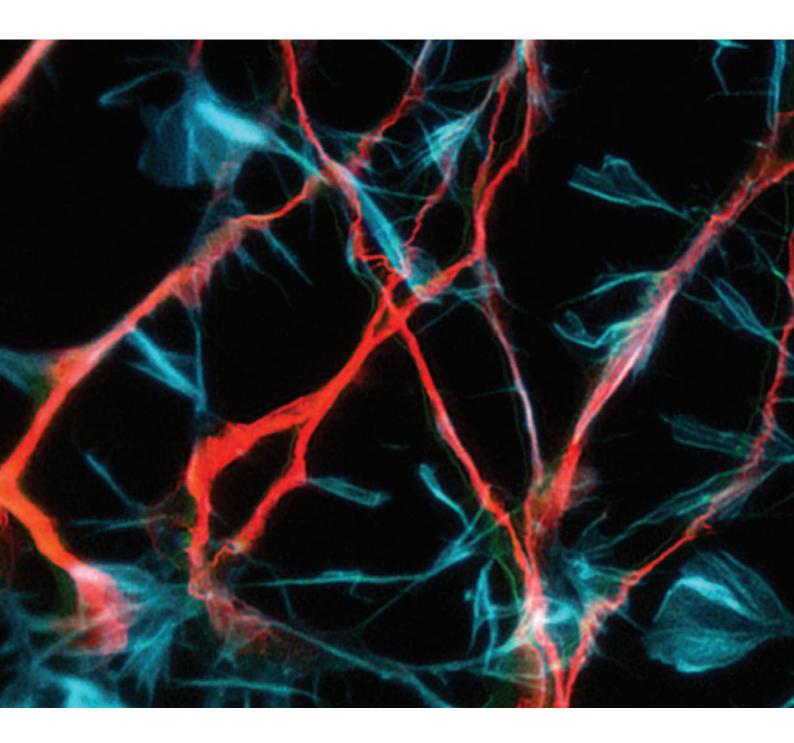
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