

Curated:

Go8 Genomic and Precision Medicine Capability 2018



Contents

Introduction	4
---------------------	----------

The Australian National University (ANU)	6
UNSW Sydney	10
The University of Adelaide	14
The University of Western Australia	18
The University of Sydney	24
The University of Melbourne	28
Monash University	34
The University of Queensland	40

Attachment A: Go8 key personnel	46
--	-----------

Introduction

A Go8 Genomics and Precision Medicine Collaboration and Commercialisation Summit with 127 participants was held in Sydney in July 2018.

A range of leading researchers provided the scientific basis for the Summit and extended discussion to the practicalities of advancing genomics and precision medicine outcomes. Representatives of venture capital and investment organisations, as well as key pharmaceutical companies also provided their perspectives.

It was a day of robust and fruitful discussions about how best to take this vital area of the Go8's research forward for the benefit not just of Australia's population but that of the global community.

At the Summit it was decided that one of the many positive outcomes should be the release of a Go8 capability statement on Genomics and Precision Medicine. This publication is the result.

Developing the publication was an exercise that, even more than the Summit, displayed the strength of Go8 research in the field.

Australia has a Government determined to prioritise Genomics and Precision Medicine and the nation has the Medical Research Future Fund, the Australian Genomics Health Alliance, the Biomedical Translation Fund, the Genomics Health Futures Mission as foundations for long-term political support.

Go8 universities apply genomics and precision medicine capability and techniques in a variety of ways to their research, collaborations and translational practices across many fields, as such I hope this publication becomes a worthy "go-to" manual of the work in this area by Go8 members.

Importantly I would like to point to one key component of Go8's capability and capacity – the ability to lead and share cutting-edge research infrastructure facilities instrumental to genomics and precision medicine advances today.

Go8 universities host major national facilities, such as the National Computational Infrastructure including one of the country's two national research supercomputers, the Australian Phenomics Network, the Ramaciotti Centre for Genomics; and are partners or host major nodes in other national research infrastructure such as BioPlatforms Australia; and provide other key facilities such as the Monash Biomedical Proteomics Facility and the South Australian Genome Editing (SAGE) Facility.

We have so much to contribute.



Vicki Thomson
Go8 Chief Executive



Australia has a Government determined to prioritise Genomics and Precision Medicine and the nation has the Medical Research Future Fund, the Australian Genomics Health Alliance, the Biomedical Translation Fund, the Genomics Health Futures Mission as foundations for long-term political support.

The Australian National University (ANU)

ANU contact point:

Dr Robin Fieldhouse

Strategic Projects and
Partnerships, Life Sciences

T: +61 2 6125 1326

E: Robin.Fieldhouse@anu.edu.au

Skills and capability

ANU has core genomics research capabilities of global standing covering basic research through to translation, consisting of: cutting-edge genomic and biological science; nation-leading RNA and chromatin expertise; Australia's most advanced high throughput functional genomics capabilities; and leading CRISPR-based genotype-to-phenotype expertise for modelling genetic mutations identified by precision medicine.

Research strengths

The ANU has international recognised leaders in personalised immunology:

- Leadership of the Genomics Australia National Genetics Immunology Flagship
- Centre for Personalised Immunology (CPI) established in 2013 (NHMRC Centre of Research Excellence)
- Recognised for novel discovery 'looping back' to inform patient diagnosis and treatment, with successful patient diagnosis and treatment in a significant number of cases





-
- Developed clinician/cohort networks in 20+ countries throughout Europe, the Middle East, India and China
 - Strategic alliance formed with Renji Hospital, Shanghai Jiaotong University (China-Australia Centre for Personalised Immunology).

The ANU also has shared clinical genomics strengths partnering with ACT Health

- Canberra Clinical Genomics (CCG) is a joint ANU/ACT Government initiative established with \$7.3 million of ACT Government funding in 2016
- CCG is key a component of the broader Genomics Australia network
- ANU performs sample preparation, sequencing and bioinformatics analysis
- ANU and The Canberra Hospital are also founding members in the \$50 million Australian Genomic Cancer Medicine Program (sequencing patients with rare cancers and advanced stage cancers to identify more effective treatments).

The university makes major investments in precision medicine

- Our Health in Our Hands” (OHIOH) – a \$10 million ANU investment over five years aims to investigate and demonstrate how personalised medicine can address major global health challenges (diabetes and MS)
- ‘WearOptimo’ – a concept of ‘microwearables’ for episodic and continuous health monitoring, is a collaboration between ANU, WearOptimo, the Queensland Government, Johnson & Johnson Innovation and the Australian National Fabrication Facility (ANFF).

Key ANU personnel, with expertise in the following areas, are listed at **Attachment A**.

- Genome informatics
- Phenomics
- Genome Editing and Genetics of Host-Pathogen Interactions
- OHIOH, Personalised Immunology
- Indigenous Genomics
- Genetics and Infectious Diseases
- Cancer Biology & Therapeutics
- Clear Vision Research Lab

The Australian National University (ANU)

-
- RNA Biology
 - Epidemiology and Population Health
 - Quantitative and Computational Biology
 - Epidemiology and Population Health
 - Genome Science
 - OHIOH, phenotype monitoring
 - Personalised Immunology
 - Social studies of biomedicine and the life sciences.

Research infrastructure

ANU has unique unparalleled national infrastructure.

- National Collaborative Research Infrastructure Strategy (NCRIS) National Computational Infrastructure (NCI)
- NCRIS Australian Phenomics Facility (APF)
- NHMRC Centre for Personalised Immunology (CPI)
- National Centre for Indigenous Genomics (NCIG) (and collection)
- Australian Cancer Research Foundation (ACRF) Biomolecular Research Facility (BRF)

-
- ACRF Target and Drug Discovery Platform for high throughput screening
 - ANU Biological and Data Science Institute (B&DSI)
 - National Centre for Epidemiology and Population Health (NCEPH).

Collaborations

ANU has a large-scale collaboration with Renji Hospital, Shanghai Jiaotong University, representing the China-Australia Centre for Personalised Immunology (CPI). This is focussed on scaling and translating the capability in the Australian node of CPI to a major Chinese context and leveraging some of the national infrastructure at the ANU (NCI). One benefit of this partnership is a massive expansion of the reference genome. Other related collaborations involve developed clinician/cohort networks in 20+ countries throughout Europe, the Middle East and India, as well as China.

ANU has a strong relationship with ACT Health, which operates on multiple levels and through multiple programs in education, research and health services, featuring many joint appointments and investments. There are multiple collaborations in progress,

two of which relate specifically to Precision Medicine/Genomics: Canberra Clinical Genomics (CCG) and the Our Health in Our Hands (OHIOH) project.

- CCG formed in 2016 relates to the translation of discovery genomics to practical application in the clinic.
- OHIOH represents a major ANU investment to investigate and demonstrate how genomics

ANU undertakes forward genetic screening work for a number of clients/partners, the most notable of which is Genentech. In this relationship which has developed over the last decade, ANU provides Genentech with access to unique genetic screening capabilities, in turn benefitting from Genentech's ability to help address health challenges through development of transformative medicines and

ANU has a large-scale collaboration with Renji Hospital, Shanghai Jiaotong University, representing the China-Australia Centre for Personalised Immunology (CPI).

in combination with precision phenotyping, data science and implementation science can be applied to address major global health challenges. In the first instance, target diseases are diabetes and multiple sclerosis. The OHIOH project includes a collaboration with CSIRO.

ANU naturally also collaborates closely with CSIRO in the genomics and precision medicine domain. Two exemplars of this are the OHIOH project above and the Biological Data Sciences Institute.

commercialisation of new discoveries on a global scale. The ANU-Genentech association has recently been expanded into neuroscience and immune-oncology, in addition to sepsis, and is expected to continue.

ANU has a number of other collaborative projects of a commercial nature focused on development of biomarkers as companion diagnostics for use in precision medicine, informed by genomics and transcriptomics. Illustrative examples include ophthalmology, diabetes and multiple sclerosis.

UNSW Sydney

UNSW Sydney contact point:

Mr Warwick Dawson,
Director, Knowledge Exchange

Division of Enterprise

E: knowledge.exchange@unsw.edu.au

Skills and capability

The University of New South Wales has a long-standing commitment in genomics and personalised medicine and is home to two key facilities: the Ramaciotti Centre for Genomics and the Microbiome Research Centre (MRC). Research projects relating to genomics conducted at UNSW range from sequencing koalas to precision medicine for blood cancer. The MRC uses powerful multi-omic approaches to define microbial signatures relevant to diagnosis, prognosis and prevention of disease.

Research strengths

UNSW Sydney has key research strengths in:

- genetic susceptibility to ovarian cancer including in the identification of germline common and rare variants associated with risk of ovarian cancer
- identification of prognostic markers for ovarian cancer¹



¹ Professor Susan Ramus leads the work on ovarian cancer.



-
- inherited blood diseases, with gene editing techniques representing a new approach to treating sickle cell anemia and related hemoglobinopathies²
 - study of healthy blood stem cells and leukaemic cells at genome-scale and in single cells to assist with understanding and management of blood stem cell disorders³

-
- the genetic basis of autism spectrum disorders⁵.

Details of the UNSW Sydney researchers involved and their foci / projects are at **Attachment A**.

The Microbiome Research Centre (MRC)⁶ is UNSW's recently established comprehensive world-

The University of New South Wales has a long-standing commitment in genomics and personalised medicine and is home to two key facilities; the Ramaciotti Centre for Genomics and the Microbiome Research Centre (MRC).

-
- application of genomics and single cell technologies in the field of immunology, using experimental techniques and bioinformatics research in virology, immunology, and epidemiology of infectious diseases, with current focus on analysis to revolutionise treatment of cancer, autoimmunity, and infectious diseases⁴

class microbiome-focused research centre. It is embedded within the grounds of the St George and Sutherland Hospitals, a major clinical campus in southern Sydney, and is led by clinicians and scientists who use state of the art science to answer important health questions relevant to Australia and the world. It comprises five research themes:

² Professor Merlin Crossley's lab investigates inherited blood diseases.

³ Professor John Pimanda's molecular and cell biology lab conducts this research.

⁴ Dr Fabio Luciani's lab leads the work in the field of immunology.

⁵ Dr Irina Voineagu leads the work on autism spectrum disorders.

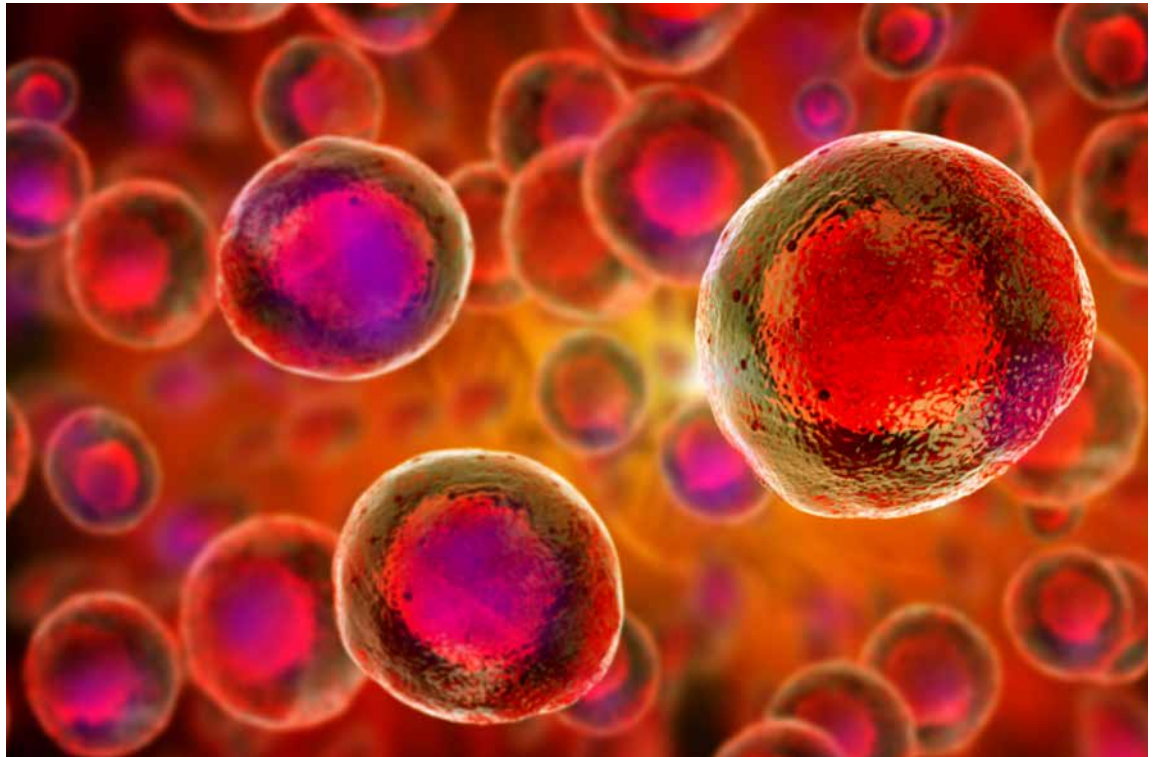
⁶ Professor Emad M El Omar is the Director of the MRC.

cancer, women's/children's health, mental health/neuroscience, critical care and infection, inflammation & immunity. Powerful multi-omic approaches are employed to define microbial signatures relevant to diagnosis, prognosis and prevention of disease. It has the potential to deliver large clinical trials through an established network of local, national and international collaborations.

The MRC will deliver personalised and precision medicine tools that are patentable and commercially attractive.

Research infrastructure

The Ramaciotti Centre for Genomics⁷ is a national infrastructure facility and a focus for the development and application of genomics in Australia.



⁷ Professor Marc Wilkins is the Director of the Ramaciotti Centre for Genomics.

It was established in 1999 with an ARC Research Infrastructure Equipment and Facilities (RIEF) grant and the Clive and Vera Ramaciotti Millennium Award (2000). It is a Centre of The University of New South Wales and sits within the Faculty of Science. The facility is located in the Biosciences Building, Kensington Campus, UNSW Sydney. The purpose of the Centre is to deliver internationally competitive genomic services by providing clients with access to enabling technology and services of the highest quality. The Centre's technology suite includes long and short-read next-generation sequencing, single-cell analysis, microarrays and capillary sequencing. The objectives of the Centre are to provide:

- users with enabling technology and services, to facilitate internationally competitive research
- genomics and related services of the highest possible quality
- support for bespoke and niche projects
- services at competitive rates.

Collaborations

The Microbiome Research Centre is a key collaboration with the St George and Sutherland Medical Research Foundation (SSMRF) supported by the Australian Federal Government, NSW State Government and South Eastern Sydney Local Health District.

The Ramaciotti Centre is governed by a number of key stakeholders including The University of Sydney, Macquarie University, The University of Technology Sydney, The University of Newcastle, the Victor Chang Cardiac Research Institute, the Garvan Institute of Medical Research and the Kolling Institute. UNSW is affiliated with the Garvan Institute of Medical Research and Kinghorn Centre for Clinical Genomics.

Other key collaborations include with:

- Ovarian Cancer Association Consortium (OCAC)
- Ovarian Tumor Tissue Analysis (OTTA) Consortium
- Autism CRC Genomics program

The University of Adelaide

The University of Adelaide contact point:

Professor Andrew Zannettino,
Professor of Experimental
Haematology

T: +61 8 8128 4390

E: andrew.zannettino@adelaide.edu.au

Skills and capability

The University of Adelaide's research expertise in health and medicine encompasses a broad spectrum of research interests, from laboratory-based biochemical research to clinical research and the study of broader populations.

The Faculty of Health & Medical Sciences has a prestigious reputation for outstanding theoretical and applied research with formal ties to the South Australian teaching hospitals, research institutes and a broad range of clinical practices.

These ties open opportunities for research collaborations across laboratory, clinical and community health settings, and allow the University to support important cross functional research needs across academia, industry and government.

The University has established genomics services and bioinformatics platforms to support its genomics, pharmacogenomics and precision medicine research in cancer, reproductive health and neurological disease.



Research strengths

The University of Adelaide's research strengths include the following:

- A comprehensive translational research program focused on understanding the determinants of response and resistance to therapy in leukaemia – with a particular focus on chronic myeloid leukaemia (CML)⁸
- Genetics and genomics of intellectual disabilities, autisms, epilepsies and cerebral palsies, with to date discovery or contribution to the discovery of more than 200 individual childhood onset neurodevelopmental disease genes⁹
- Cerebral Palsy Research, via leadership of the Australian Collaborative Cerebral Palsy Research Group which has the world's largest DNA biobank and clinical data bank from Cerebral Palsy (CP) families. The team leads in demonstrating there is genomic basis to many cases of CP using genomic sequencing and epigenetic studies¹⁰
- Use of CRISPR preclinical mouse models in delivering unprecedented insight into the pathology underpinning a host of genetic diseases including cancer, epilepsy and intellectual disability, including recently to develop novel CRISPR therapy strategies for Duchenne Muscular Dystrophy¹¹
- Next-generation sequencing of newly diagnosed and relapsed/refractory Acute Lymphoblastic Leukaemia (ALL) patients of all ages from around Australia, through the Australian Genomics Health Alliance ALL flagship¹²

⁸ Professor Timothy Hughes leads the work on leukaemia.

⁹ Professor Josef Gecz heads the Neurogenetics Research Program (NEURO).

¹⁰ Led by Emeritus Professor Alastair MacLennan and Professor Josef Gecz.

¹¹ Professor Paul Thomas is a leader in CRISPR/Cas9 genome editing.

¹² Professor Deborah White's group leads this work.

- Research in predicting in early pregnancy which women are at risk of developing the four major complications of pregnancy that afflict 25 per cent of first pregnancies in Australia and which are life-threatening to the mother and/or baby in about six per cent of pregnancies, including by using genomic, transcriptomic and epigenomic analyses to develop methods to monitor placental health non-invasively in real time right across gestation¹³
- Pharmacogenomics research combining pharmacology and genomics expertise to explain why medicines don't work or are toxic in certain patients, with current work on developing markers for safer and more effective treatment in pain and psychiatry, infection, cancer and transplantation, and in researching indigenous Australians¹⁴.

Details of key University of Adelaide researchers and their work in the above areas of focus are at **Attachment A**.

Research infrastructure

The University of Adelaide has the world's largest DNA biobank and clinical data bank from Cerebral Palsy families, with an international CP genomic databank to be based at The University of Adelaide.

The University of Adelaide also co-hosts with the South Australian Health and Medical Research Institute (SAHMRI) the South Australian Genome Editing (SAGE) Facility which is supported through the NCRIS-funded Australian Phenomics Network.

Collaborations

The University has formal ties to the South Australian teaching hospitals, research institutes and a broad range of clinical practices. It has strong collaborations with Pharma and international trial groups, in transforming outcomes for chronic myeloid leukaemia (CML) patients globally.

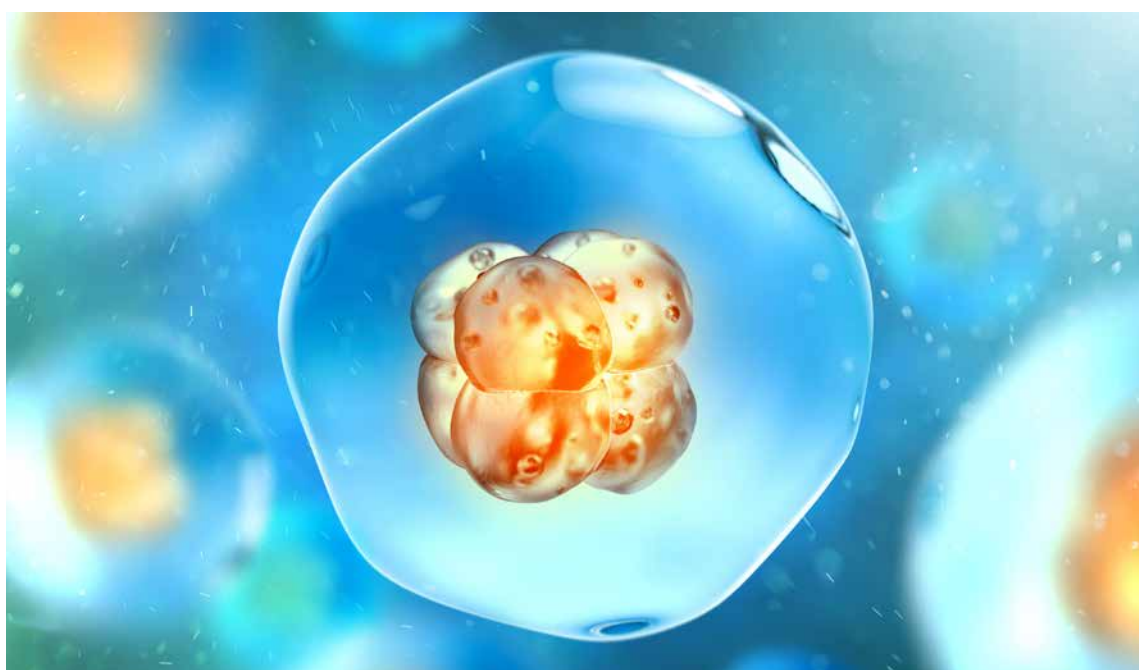
¹³ Professor Claire Roberts and Professor Gus Dekker lead the Placental Development Group.

¹⁴ Professor Andrew Somogyi and Dr Dan Barrett are key personnel involved.

The University's Neurogenetics Research Program (NEURO) team has major national and international linkages with universities and health services. In epilepsies NEURO is the genomics expert of current NHMRC Epilepsy Program grant (CIA Berkovic from The University of Melbourne). In intellectual disabilities NEURO is the founding member of the Autism Spectrum Intellectual Disability (ASID) Consortium currently led by Prof Eichler from Seattle, USA, Leader in NHMRC funded CRE in Neurocognitive disorders as well as a long-term research collaborator (>25 years) of the Genetics of Learning Disability Service (GOLD) of NSW.

The University also has collaborations in pharmacogenetics with UNSW, The University of Melbourne, Monash University, The University of Auckland, Singapore KK Women's and Children's Hospital, and the European Pharmacogenetics of Opioids Study, La Trobe University and the University of Papua New Guinea.

Additionally, the University has links to and contributes to the Australian Genomics Health Alliance, Australian Functional Genomics, the International Clinical Pharmacogenetics Implementation Consortium and the RCPA Intercollegiate Pharmacogenetics Working Group.



The University of Western Australia

The University of Western Australia contact point:

Dr Samantha South,
Senior Commercialisation
Officer (Life Sciences)

Research Development
and Innovation

T: +61 6488 3905

M: +61 42 111 7241

E: samantha.south@uwa.edu.au

Skills and capability

The University of Western Australia is one of Australia's leading universities and has an international reputation for excellence in teaching, learning and research. In 2018, the University sits at 93 on the Academic Ranking of World Universities produced by Shanghai Jiao Tong University.

Harry Perkins Institute of Medical Research conducts innovative research in oncology, genetic, cardiovascular and metabolic disease that translates into lasting health benefits.

Research strengths

The University of Western Australia has research strengths in the following. Key UWA personnel, with expertise in the following areas, are listed at **Attachment A**.



Harry Perkins Institute of Medical Research conducts innovative research in oncology, genetic, cardiovascular and metabolic disease that translates into lasting health benefits.

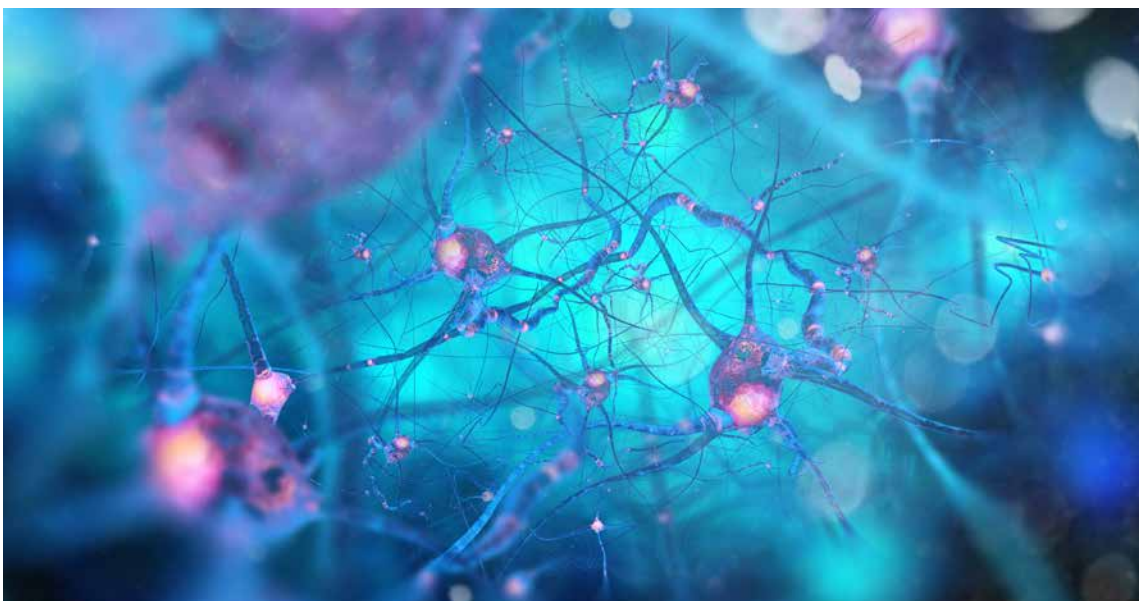
Neurogenetics diseases¹⁵

Disease Gene Discovery:

- involvement in the discovery of more than 30 human disease genes. Many of these cause severe muscle or nerve diseases prior to or at the time of birth
- research in adult peripheral nerve and muscle diseases and inherited neurodegenerative disorders
- each disease gene discovery leads to precision medicine diagnosis for patients around the world affected by pathogenic variants in the gene.

Development of improved diagnostics:

- developed and implemented next generation sequencing diagnostics for neurogenetic diseases in partnership with the Department of Diagnostic Genomics, Department of Health Western Australian PathWest laboratories, forming a national referral centre for the molecular diagnosis of these diseases and significant increase in precision medicine diagnosis
- developing new diagnostic tools to enhance success rates in the diagnosis of patients with neurogenetic diseases.



¹⁵ The Neurogenetic Diseases Group is led by Group Leader Professor Nigel G Laing AO.

Development of therapies:

- researching precision medicine genetic therapies for specific genetic muscle diseases, including diseases such as the skeletal muscle actin diseases (first identified by the Group).

Prevention of genetic disease:

- extended advocacy for both newborn and carrier screening
- involvement in Mackenzie's Mission, the \$20 million Medical Research Future Fund research project on

implementation of carrier screening in Australia: one of the largest precision medicine research projects in Australia.

Systems Biology and Genomics¹⁶

This includes in single cell profiling of tumours – tumour microenvironment, receptor-ligand signaling networks, biomarkers, precision oncology; disease gene prioritization – tissue specific expression and tissue specific phenotypes; copy number variation; and long non-coding RNA function.



¹⁶ The Systems Biology and Genomics Lab Group Leader is Professor Alistair Forrest.

Characterizing and editing the epigenome¹⁷

This includes epigenome analysis in health and disease, including regenerative medicine, neurodevelopment, and neurological disorders, and the use of precision tools for deliberately editing the epigenome in human cells.

Centre for Genetic Origins of Health and Disease (GOHaD)¹⁸

GoHaD is home to a multidisciplinary team of statistical and molecular geneticists, bioinformaticians, computer programmers and epidemiologists. The team collaborates across disciplines and organisations to determine the genetic, epigenetic and environmental contributions to risk of common disease, including cardiovascular disease, neuropsychiatric disorders, pregnancy disorders and several cancers including melanoma, mesothelioma, sarcoma, colorectal cancer and breast cancer.

Translational Cancer Pathology¹⁹

Work focuses on translating scientific discoveries into diagnostic pathology for clinical application. This includes:

- predicting progression in myeloproliferative neoplasms
- blood-based monitoring of malignancies (“liquid biopsy”) using circulating tumour cells by imaging flow cytometry, cfDNA, exosomes, tumour educated-platelets
- imaging flow cytometry for the detection of chromosomal abnormalities in immunophenotyped cells (“immuno-flowFISH”: UWA patented technology)
- a commercial partnership with Sysmex.

¹⁷ The Group Leader for Characterising and Editing the Epigenome is Dr Ryan Lister.

¹⁸ GoHAD Group Leader is Professor Eric Moses.

¹⁹ Translational Cancer Pathology Group Leader is Professor Wendy Erber.

Genetic Eye Diseases²⁰

All major eye diseases have a strong genetic component (age-related macular degeneration, primary open-angle glaucoma, Myopia, Cat, DR) and the leading cause of blindness in working age adults is inherited retinal disease.

The University of Western Australia Genetic Eye Diseases lab focusses on discovery of genetic and environmental factors associated with eye disease, in particular myopia and glaucoma, through the NHMRC funded Centre of Research Excellence “From discovery to therapy in genetic eye diseases”.

The lab collaborates with the genetic eye disease research teams in every state. Major collaborations are with the Consortium for Refractive Error and Myopia (CREAM) and the International Glaucoma Genetics Consortium (IGGC). The team will also participate in a nationwide NHMRC Program grant for Glaucoma Genetics in 2019.

Immuno-oncology²¹

Work entails identification of biomarkers and new drug targets in immuno-oncology, and involves:

- unique animal models that allow the mapping of the biological events that govern the response to immunotherapy
- systems biology platform to pinpoint druggable targets (team of lab scientists, computational biologists and mathematicians)
- in vivo validation in animals and patient samples
- excellent track record in investigator-initiated clinical trials extensive experience with in vivo immuno-monitoring
- interest in combination of standard of care (surgery, radiotherapy, chemotherapy) and immunotherapy
- neoantigen discovery and T-cell Receptor (TCR) characterization
- a commercial partnership with Douglas Pharmaceuticals.

²⁰ Genetic Eye Diseases Group Leader is Professor David Mackey.

²¹ Immuno-oncology Group Leader is Associate Professor Joost Lesterhuis.

National Centre for Asbestos Related Diseases²²

The Centre provides basic, translational and clinical research in asbestos-related diseases, including:

- Molecular biology and genetics of mesothelioma; biomarker research, small molecule drug development, clinical trials (phase 1–3).
- animal models of carcinogenesis and genetic predisposition

Collaborations

The University of Western Australia's key partnerships and alliances are in or with:

- Mackenzie's Mission
- NHMRC Program grant for Glaucoma Genetics
- The Consortium for Refractive Error and Myopia (CREAM)

The University of Western Australia Genetic Eye Diseases lab focusses on discovery of genetic and environmental factors associated with eye disease ... The team will also participate in a nationwide NHMRC Program grant for Glaucoma Genetics in 2019.

Research infrastructure

The University of Western Australia collaborates with the NCRIS Australia Phenomics Network.

- The International Glaucoma Genetics Consortium (IGGC).
- The Single Cell Cancer Genomics Consortium
- The Australian Phenomics Network.

²² National Centre for Asbestos Related Diseases Group Leader is Professor Anna Nowak.

The University of Sydney

The University of Sydney contact point:

Ms Jessica Keath,
Manager

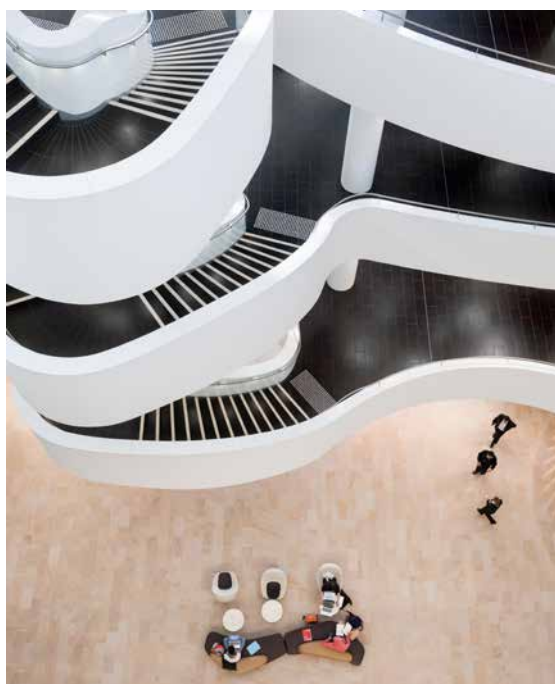
Research Development
(Health & Medical Research),
Research Portfolio,
Office of Deputy Vice
Chancellor Research

T: +61 2 8627 6383

E: jessica.keath@sydney.edu.au

Skills and capability

The University of Sydney has leading genomics capability in the areas of constitutional diseases²³, cancer²⁴ and infectious diseases²⁵. It is important to the University that our understanding of genomics and its integration into the healthcare system connects with other kinds of 'omics' (transcriptomics, proteomics and metabolomics among others), data analytics, health ethics and psychosocial research, all of which are needed to realise the full potential of precision medicine in Australia.



The University of Sydney has leading genomics capability in the areas of constitutional diseases⁹, cancer¹⁰ and infectious diseases¹¹.

²³ Professor Robyn Jamieson, Professor Christopher Semsarian, Professor Carolyn Sue, Professor Ian Alexander.

²⁴ Professor Roger Reddel, Professor Graham Mann, Professor Robyn Ward.

²⁵ Professor Edward Holmes, Professor Jonathan Iredell, Professor Tania Sorrell, Professor Vitali Sintchenko.



Research strengths

The University's key translational strengths in these areas are in translational and therapeutic genomics (e.g. gene therapy, vectorology) cancer genomics and proteomics (e.g. Sydney Cancer Research Network) and precision medicine for infectious diseases (e.g. pathogen discovery, mycology genomics and precision public health, evolution and emergence of infectious diseases, transmissible antimicrobial resistance and rapid response diagnostics).

Key expertise is in the areas of:

- translational genomic medicine for retinal and developmental eye diseases (Professor Robyn Jamieson)
- genetic therapeutic approaches in retinal organoids, aimed towards treatment for these blinding eye conditions
- clinical and genetic aspects of inherited heart diseases (Professor Chris Semsarian)
- Parkinson's disease and mitochondrial disorders, using genomic precision medicine to identify gene mutations in mitochondrial disease (Professor Carolyn Sue).



Further details on The University of Sydney researchers leading research in the above are **Attachment A**, along with details of other key researchers and their research areas.

Research infrastructure

The University of Sydney has a wide range of relevant research infrastructure.

- ACRF International Centre for the Proteome of Human Cancer (ProCan): ProCan provides high-throughput cancer proteomics to address the current deficit in the multi-omic analysis of cancer.

parallel computing and/or computer clusters for advanced computing tasks to enable sophisticated genomics data analysis.

- Sydney Cytometry: Specialist facilities for cell sorting with flow, mass and image cytometric platforms delivering rapid throughput and the ability to monitor large numbers of parameters.
- Sydney Imaging: Biomedical imaging research facility with a focus on clinical, preclinical and hybrid theatre capability. The University's clinical and preclinical

Sydney Health Partners (SHP), one of the first four Advanced Health Research and Translation Centres in Australia, recognised by the NHMRC in 2015 for being a world leader in translational research.

- Bioinformatics Core Facility: This facility is developing specialist analytic methods for assessing proteomic and metabolomic data sets to support precision medicine.
- High Performance Computing (HPC): The University of Sydney's HPC makes use of supercomputers,

capabilities are focussed around MRI and X-ray technologies and data workflows position imaging as a key enabler of precision medicine.

- Sydney Mass Spectrometry: Outstanding facilities for genomics, proteomics and lipidomics, with the capacity to conduct these assays

at a clinical scale with a precision medicine agenda.

- Western Sydney Genetics Program: These facilities and services are research active in genomics and precision medicine, in addition to their service roles, and are led by University of Sydney Clinical Academics or Titleholders:
 - » Sydney Genome Diagnostic Laboratories
 - » Biochemical Genetics Laboratory
 - » Newborn Screening Laboratory
 - » Clinical Genetics Service
 - » Genetic Metabolic Diseases Service.

Collaborations

The University of Sydney's key partnerships and alliances include in:

- The Australian Genomics Health Alliance (AGHA) is an NHMRC funded alliance²⁶.
- Sydney Health Partners (SHP), one of the first four Advanced Health Research and Translation Centres in Australia, recognised by the NHMRC in 2015 for being a world leader in translational research. The University of Sydney is a founding partner of SHP²⁷.
- Paediatrico, a comprehensive collaborative network of researchers and clinicians across NSW with the goal of building scale in translational paediatric research and includes the key research theme of Applied and Translational Genomics. University of Sydney researchers (included in **Attachment A**) at The Children's Hospital at Westmead and Children's Medical Research Institute are founding members.

²⁶ University of Sydney affiliated AGHA investigators are: Associate Professor Bruce Bennetts, Professor Chris Cowell, Associate Professor Sandra Cooper, Professor Robyn Jamieson, Associate Professor Kristi Jones, Associate Professor Ainsley Newson, Professor Roger Reddel, Professor Christopher Semsarian, Professor Carolyn Sue and Professor Robyn Ward.

²⁷ Professor Robyn Jamieson leads SHP's Genomics Cross Cutting Theme.

The University of Melbourne

The University of Melbourne contact point:

Dr Heather St John,
Director

MRFF Initiatives,
Research Innovation
and Commercialisation

T: +61 3 8344 7172

E: hstjohn@unimelb.edu.au

Skills and capability

As a globally engaged, comprehensive research institution, The University of Melbourne works closely with its Melbourne Biomedical Precinct (MBP) partners to continually challenge the status quo in research and clinical benchmarks. With a significant emphasis on progressing cross-disciplinary, translational research in emerging fields of endeavour, such as Precision Medicine, The University of Melbourne and its clinical partners focus on improved patient diagnoses, targeted drug discovery and accelerated clinical validation of new drug candidates, to ensure excellence in research while striving to deliver impact to patients and society on a global scale.

The Melbourne Biomedical Precinct is Australia's, and one of the world's, leading biomedical precincts. The 30 precinct partners and some 10000 researchers are engaged in breakthrough biomedical and healthcare research, particularly in child and adolescent health, cancer, mental health and neurosciences, infectious diseases and healthy ageing.





The University benefits from the strong collaborative frameworks established in the Precinct to integrate research and clinical delivery for the benefit of Victorians, notably the Melbourne Academic Centre for Health, Victorian Comprehensive Cancer Centre and Victorian Clinical Genetics Service. Through MACH's 10 partner hospitals, our clinical partners reach over 2.3 million people – 49 per cent of Melbourne, 37 per cent of Victoria, and 10 per cent of Australia's population. This connectivity creates an opportunity to rapidly test research outcomes, including new therapeutic interventions, and deliver broad impact.

Research strengths

Research groups across the University and embedded in hospital departments are focused on conducting world-class research to advance the field of Genomics and Precision Medicine particularly in Precision Oncology, Microbial Genomics, Immunogenetics, Genetic Diagnostics of Epilepsy, Stem Cell Genomics, Population and Statistical Genomics.

Testament to the ground-breaking research undertaken in precision medicine by University researchers, in the last six years over 3700 publications related to precision medicine were published, with a field-weighted citation impact of 3.5. Over 40 per cent of the University's precision medicine publications are in the top 10 per cent of the most cited publications world-wide and over 55 per cent of precision medicine publications are published in the top 10 per cent of journals in the world.

Specific expertise is in:

- **Precision Oncology**²⁸ – Cancer detection and surveillance, monitoring drug response and studying evolution of chemo-resistance, using liquid biopsies
- **Microbial/pathogen genomics**²⁹ – Use of high-throughput platforms to sequence the complete genomes of drug-resistant and drug-susceptible bacterial pathogens, to investigate underlying mechanisms and impact

²⁸ Precision Oncology Leader: Professor Sean Grimmond.

²⁹ Microbial/pathogen genomics Leaders: Professor Kathryn North, Associate Professor Kathryn Holt.

-
- **Genetic epidemiology**³⁰ – Application of statistical methodology for analysing twin and family data, addressing the genetic and environmental aetiology of diseases and health
 - **Genetic diagnostics of epilepsy**³¹ – Clinical and molecular genetic studies are conducted by studying individuals and families with a history of seizures using cutting edge research methods and technologies
 - **Statistical Genomics**³² – Developing methods for studies of population structure and immunogenetics
 - **Immunogenetics**³³ – Molecular genetic studies of immune system function and dysfunction
 - **Clinical Genetics**³⁴ – Developing and validating new methods of cancer risk prediction

In addition, Certa Therapeutics, founded by Professor Darren Kelly, with its laboratories based at The University of Melbourne, uses genetic analysis to identify the patients that will develop kidney fibrosis – the precursor to end-stage kidney failure – and respond to pre-emptive treatment. The precision medicine revolution will save lives and spare patients from dialysis by using Certa's treatments to block a receptor that is a key driver of the fibrosis. The underlying technology originated from The University of Melbourne, Bio21 and St Vincent's Institute for Medical Research.

The University of Melbourne is also a partner in the Australian Research Council (ARC) Training Centre for Personalised Therapeutic Technologies which focuses on skill development in precision medicine. Professor Alastair Stewart and colleagues received over \$3.2 million for an Industrial

30 Genetic epidemiology Leader: Professor John Hopper.

31 Genetic diagnostics of epilepsy Leaders: Professor Ingrid Scheffer and Professor Sam Berkovic.

32 Statistical Genomics Leaders: Professor David Balding and Associate Professor Stephen Leslie.

33 Immunogenetics Leader: Professor James McCluskey.

34 Clinical Genetics Leader: Professor Ingrid Winship.

Transformation Training Centre based collaboration between The University of Melbourne, Monash University, The University of Western Australia and 15 partner organisations. Based in the University's Department of Pharmacology and Therapeutics, the Centre focuses on training biomedical researchers to enable early benefit from the transformative impacts of cell/organ-on-a-chip technology. Combining microfluidics/real-time technologies and individual patients' cellular and molecular data, will enable personalised diagnostic and therapeutic decisions.

Key personnel, with expertise in the above areas, are listed at **Attachment A**.

Research infrastructure

Melbourne Clinical Genomics Platform

The Melbourne Clinical Genomics Platform makes the most of the genomic capabilities of the University of Melbourne Centre for Cancer Research (UMCCR) located within the Victorian Comprehensive Cancer Centre. The technologies and the expertise of supporting staff are readily

accessible to researchers, clinician and students across the Melbourne Biomedical Precinct. Through the department of Clinical Pathology an Illumina NovaSeq 6000 and MiSeq Sequencing System is available.

Molecular Diagnostics – Our NATA compliant ISO 15189 (Medical Testing) laboratory offers specialised services to enable rapid large scale sequencing for patient monitoring and precision oncology.

Collaborations

There is a strong commitment to collaborative, strategic partnerships across the Melbourne Biomedical Precinct, and considerable investment has been made to create targeted alliances to pioneer Precision Medicine by The University of Melbourne and precinct partners. These include:

- International Cancer Genome Consortium – 2010–18 (The University of Melbourne, Walter and Eliza Hall Institute, Peter MacCallum Cancer Centre – \$28 million): This NHMRC supported program pioneered genome sequencing in Australia, studying the root causes and druggable targets in >1000

patients suffering from our most challenging cancers: pancreatic, ovarian and melanoma.

- Melbourne Genomics Health Alliance – 2014–2019 (The University of Melbourne, Walter and Eliza Hall Institute, Peter MacCallum Cancer Centre, Murdoch Children’s Research Institute – \$25 million): Supported by State Government, the MGHA has demonstrated clinical utility of exome sequencing as primary testing for RUGD.
- Australian Genomics – 2016–2020 (The University of Melbourne, Walter and Eliza Hall Institute, Peter MacCallum Cancer Centre, Murdoch Children’s Research Institute- \$25 million): Built on MGHA, this NHMRC supported program has leveraged similar state-based programs focused on delivery of molecular testing for rare and undiagnosed genetic diseases, panel/RNA sequencing and exome/who-genome sequencing based analysis of cancer and acute care genomic diagnostics for Neonatal intensive care unit (NICU)/Paediatric intensive care unit (PICU) patients.
- Global Alliance for Genomics and Health (Murdoch Children’s Research Institute, The University of Melbourne): GA4GH is a consortium of 400+ institutions (including Australian Genomics, Genomics England, Genome Canada, US Precision Medicine Initiative) in 46 countries committed to developing international standards for data sharing and global scale translation of genomic research.
- Victorian Comprehensive Cancer Centre Alliance – 2013 (The University of Melbourne, Walter and Eliza Hall Institute, Peter MacCallum Cancer Centre, Murdoch Children’s Research Institute \$20 million): Launched in 2013, the VCCC alliance aligns both research and cancer care delivery across the precinct. Through Victorian Government support, the VCCC alliance has recently commenced a strategic research program which includes flagships in precision oncology, precision-prevention and infrastructure for undertaking innovative clinical trials.

-
- Genomics Innovation Hub:
In 2016–18 The University of Melbourne, Murdoch Children’s Research Institute, Peter MacCallum Cancer Centre, Walter and Eliza Hall Institute and Australian Genomics Research Facility (AGRF), launched the GIH to attract first access to disruptive genomic technology and has enabled the establishment of 10x single cell technology, Oxford Nanopore single molecule sequencing and early access to Novaseq platform (providing large scale patient-sequencing capacity).

Medicine programs, the University has fostered close working relationships with companies such as Amazon Web Services, Intel, Microsoft, Color Genomics, and the Google Compute Engine team, to develop and deploy large scale genomic workflows.

The University has developed long-term partnerships with international pharma and biotech companies, leveraging the research, clinical expertise and infrastructure to expedite and enhance the development of improved diagnostic

... the University has fostered close working relationships with companies such as Amazon Web Services, Intel, Microsoft, Color Genomics, and the Google Compute Engine team, to develop and deploy large scale genomic workflows.

In addition, The University of Melbourne maintains strong connections with the Bertalli Trust Fund, the Li Ka Shing Foundation, the National Institutes of Health’s National Cancer Institute, and the International Cancer Genome Consortium (ICGC). Building on the University’s bioinformatics capability underpinning Genomics and Precision

and therapeutic innovations, through sponsored research programs and large-scale clinical studies. A focus on the development of synergistic relationships with multi-national strategic partners has enabled the University to focus on world-class platform development, service provision, therapeutic discovery and clinical care.

Monash University

Monash University contact point:

Dr Svetozar Kovacevic,
Director

Faculty Research Office,
Medicine, Nursing and
Health Sciences,
Monash University

E: svetozar.kovacevic@monash.edu

Skills and capability

Monash University promotes the translation of genomics and precision medicine research through Monash Partners Academic Health Research Translation Centre, an alliance of the University, hospital networks

Monash Partners collaborate closely to share expertise and infrastructure between the University's Clayton campus, the Monash Health Translation Precinct and the Alfred Health Precinct.



(Alfred Health, Monash Health, Eastern Health, Peninsula Health, Epworth Health, Cabrini Health) and medical research institutes (Hudson Institute of Medical Research, Baker Institute, Burnet Institute). Monash Partners collaborate closely to share expertise and infrastructure between the University's Clayton campus, the Monash Health Translation Precinct and the Alfred Health Precinct. A large number of clinical and research staff across Monash Partners also have adjunct status with the University.



Research strengths

Genomics and precision medicine research at Monash University is primarily carried out at two of its research institutes (Monash Biomedicine Discovery Institute and Monash Institute of Cognitive and Clinical Neurosciences) and its largest clinical schools located at the Monash Health (School of Clinical Sciences at Monash Health) and Alfred Health (School of Public Health and Preventive Medicine, Central Clinical School) Precincts.

Oncology and Cancer – Cancer is currently one of the principal foci for precision medicine at Monash. Monash has been pivotal to the establishment of large epidemiological research resources that are now being utilised to address key questions in cancer research³⁵. Highly selected groups of people are participating in studies to support the identification of heritable risk factors (genetic and epigenetic),

while large population-based samples are enabling research to further characterise risk factors to enable clinical translation of new information. With an initial focus on common cancers such as breast and prostate cancer Monash aims to improve diagnosis and risk assessment; improve early detection, targeted therapies, prognostication and disease monitoring; devise best practice guidelines to drive necessary policy change; and develop a workforce trained in anticipation for future advances in cancer research. Specific foci include:

- The Precision Oncology Program³⁶ which uses a multidisciplinary Precision Oncology Tumour Board to guide the selection and sequencing of therapies in patients whose cancer is resistant to standard treatments and provide a mechanism to integrate the considerable clinical, molecular and research expertise that supports excellence and innovation in patient care

³⁵ This work is led by Professor Melissa Southey, Chair of Precision Medicine at the School of Clinical Sciences at Monash Health.

³⁶ The Precision Oncology program is overseen by Professor Mark Shackleton, Director of Oncology at Alfred Health and Professor Gail Risbridger, Director of Research at the Monash Partners Comprehensive Cancer Consortium.

-
- Clinical haematologists³⁷ apply molecular and genomic approaches to study blood cancers. By studying cancer cell signalling, transcriptional control networks and genetic and epigenetic regulation of normal and cancer cells, they hope to identify new and improved therapies
 - Functional cancer genomics and the role of cellular signalling in cancer³⁸.

Public Health Genomics – The Public Health Genomics Program³⁹ conducts research centred around the genetic analyses of large cohorts, clinical trials and patient registries. The Program aims to address major issues faced by the field of human genetics, such as how to effectively convert large amounts of genomic sequence data into meaningful medical information, interpret and disclose genetic information

responsibly, and integrate genetic data into medical support systems. The Program was established on the foundation of collaborative projects made possible by the NIH-backed ASPREE study and ASPREE Healthy Ageing Biobank. These projects involve the genome sequencing of thousands of individuals and integration of deep longitudinal phenotyping and clinical outcome data to explore the role of genetics in healthy ageing and disease.

ASPREE is now part of national and international genomics initiatives including the Medical Genome Reference Bank led by the Garvan Institute and the Resilience Project led by the Icahn Institute at Mount Sinai School of Medicine in New York. The Public Health Genomics Program is rapidly expanding into other projects associated with the STAREE clinical trial and Monash Clinical Registries.

37 Clinical haematologists include Professor Andrew Wei, Professor Andrew Perkins and Professor David Curtis.

38 The key researcher Associate Professor Sefi Rosenbluh is the Director of the Centre of Functional Genomics at the Hudson Institute and Monash Biomedicine Discovery Institute and a Chief Investigator for the Paediatric Precision Medicine program. Associate Prof Rosenbluh is also a specialist in CRISPR loss of function screens, a powerful gene-editing technique which allows researchers to go into a sequenced genome to pinpoint the gene that is causing or affecting a disease to “edit” the gene responsible.

39 The Public Health Genomics Program was established by Dr Paul Lacaze.

Immunology and Infectious

Diseases – including targeted and global quantitative proteomics of complex biological samples, next generation proteomics, studies of

genomic and clinical markers. In Multiple Sclerosis research⁴¹, new biomarkers are developed for axonal degeneration in MS and understanding the effects of MS risk

In Multiple Sclerosis research, new biomarkers are developed for axonal degeneration in MS and understanding the effects of MS risk genes.

the immunopeptidome in health and disease, viral antigen presentation (Epstein Barr, HIV, Vaccinia), and self-antigen presentation, tolerance and autoimmunity.

Neuroscience – including in epilepsy⁴⁰ in translating research findings into the practice of personalised medicine for epilepsy. Work aims to identify novel molecular targets for treatment and prevention, and to better understand the response to treatment by identifying biomarkers for treatment outcomes – including imaging, electrophysiological,

genes. Research also occurs into the genetic factors behind conditions such as attention deficit hyperactivity disorder (ADHD)⁴².

Diabetes and Cardiovascular

Diseases – understanding the mechanism of transcriptional control in metabolic disease, especially diabetes, and using genomic techniques to identify novel therapeutic targets for vascular disease⁴³.

A summary of the key researchers involved is at **Attachment A**.

⁴⁰ Professor Patrick Kwan and Professor Terry O'Brien are leading researchers in epilepsy.

⁴¹ Professor Helmut Butzkueven focuses on Multiple Sclerosis.

⁴² Professor Mark Bellgrove.

⁴³ Professor Sam El Osta leads this work on diabetes and cardiovascular diseases.

Research infrastructure

Monash operates a network of over 30 Technology Research Platforms. These core facilities provide specialist infrastructure, services and expertise to researchers across both academia and industry. Several of these facilities are dedicated to genomics, proteomics and precision medicine:

Sequencing, High Content Screening, Microarray, Real-Time qPCR and Fluidigm Single Cell technologies.

- The Micromon platform serves discovery research by providing genomics services including Sanger and Next Generation sequencing. These services are supported by the provision of DNA and RNA

The Monash Biomedical Proteomics Facility is a mass spectrometry based analytical platform established to provide researchers access to sophisticated proteomic technologies. These technologies allow the accurate analysis of proteins from diverse sources allowing their identification, characterisation and quantification for wide ranging studies and research outcomes.

- The MHTP Medical Genomics Facility at the Monash Health Translation Precinct (MHTP) uses the latest genomic technologies to facilitate and accelerate genomic-based translational research. The facility services research institutes throughout Australia, maintaining an excellent reputation for provision of the highest quality genomic data and support. The facility offers Sanger and Next Generation

sizing and quantification, custom oligonucleotide synthesis and other services. Micromon also runs a highly reputed molecular biology course that provides training in the essential skills of recombinant DNA technology for researchers new to the discipline.

- The Monash Bioinformatics Platform is a hub for bioinformatics activities at Monash University.

It has a core group of bioinformaticians with broad expertise and are building a linked community across all of Monash and its partners.

- The Monash Biomedical Proteomics Facility is a mass spectrometry based analytical platform established to provide researchers access to sophisticated proteomic technologies. These technologies allow the accurate analysis of proteins from diverse sources allowing their identification, characterisation and quantification for wide ranging studies and research outcomes. The facility also offers high throughput screening, accurate sequencing, protein quantitation and determination of post-translational modifications.
- The Protein Production Unit has the capacity to purify large numbers of recombinant proteins for a variety of research purposes in a high-throughput manner. The unit offers expertise in the optimisation, expression and purification of recombinant proteins in a variety of expression systems.

Collaborations

Researchers in genomics and precision medicine collaborate broadly across the local and international medical research communities. Monash University's top local collaborators in this space, outside of Monash Partners, discussed earlier, are the Walter and Eliza Hall Institute of Medical Research, The University of Melbourne, Peter MacCallum Cancer Centre, The University of Sydney, The Australian National University, The University of Queensland, Murdoch CRI, Garvan Institute and QIMR. Monash researchers are also actively involved in the Australian Genomics Health Alliance on initiatives such as the Australian Functional Genomics Network and Research Flagships (Acute Care Genomics, Renal Genetics, Mitochondrial Diseases, Genetic Immunology). Monash University's top international collaborators in the field include Harvard Medical School, University College London, University of Cambridge, Karolinska Institute and JLU Giessen.

The University of Queensland

The University of Queensland contact point:

Professor David W. Burt,
Director UQ Genomics

Office of the Deputy
Vice Chancellor Research

T: +61 7 3443 1650

E: d.burt@uq.edu.au

Skills and capability

The University of Queensland has numerous research and research collaborative strengths, with capability embedded in several key institutes at the University.

Research strengths

Statistical Genomics of Complex Traits⁴⁴ – is a particular strength at The University of Queensland and in Queensland of national and international significance: Institute for Molecular Bioscience (IMB)/ Queensland Brain Institute (QBI) and UQ Diamantina Institute (UQDI)/ Faculty of Medicine (FoM). Research includes the development of novel approaches in statistical genetics and prediction of genetic risks, systems biology, and integrative genomics for the analysis of complex conditions. Areas of research include: inheritance of common diseases, neurodegenerative and other



⁴⁴ In Statistical Genomics of Complex Traits, key personnel at IMB/QBI are Professor Peter Visscher, Professor Naomi Wray, Professor Jian Yang and Professor Grant Montgomery and, at the UQ Diamantina Institute, Professor David Evans.



neurological conditions, reproductive conditions (e.g. endometriosis), association of birthweight and adult disease, osteoporosis, ankylosing spondylitis and eczema.

Functional Genomics⁴⁵ – has been a strength at UQ for more than 20 years. In recent years gene editing tools have been used to create biomedical models of human disease, including cellular systems, organoids and whole animal models (zebrafish and mice). These capabilities are established at IMB and the Australian Institute for Bioengineering and Nanotechnology (AIBN).

Single Cell Genomics⁴⁶ – is a recent technological advance in genomics and creates new opportunities for high resolution analysis of cell populations in normal and disease conditions, such as development and cancer. At IMB, together with collaborators at The University of Sydney and The University of

Melbourne have developed the high throughput technologies of single cell gene expression assays, but most significantly, matched these by novel bioinformatics solutions to analyses such large, complex datasets. Recently, the techniques of single cell genomics and gene editing have been combined allowing the functional effects of 100s of defined mutations to be tested in cellular gene expression systems.

Infectious Disease Genomics⁴⁷ – at the Australian Infectious Disease Research Centre, a major collaboration between UQ Faculties of Medicine and Science has a particular interest in using advanced sequencing technologies (short, long and ultra-long molecule sequencing) combined with novel bioinformatics, to track hospital infections with a view to identify the source, identity, and antimicrobial resistance of new infections.

45 In Functional Genomics, key personnel are Dr Nathan Palpant, Associate Professor Ben Hogan and Professor Peter Koopman at IMB and Professor Ernst Wolvetang at AIBN.

46 In Single Cell Genomics, key personnel are Dr Sam Lukowski and Dr Quan Nguyen at IMB.

47 In Infectious Diseases Genomics, key personnel are Professor David Paterson, Professor Mark Walker, Professor Mark Schembri and Associate Professor Scott Beatson at the Australian Infectious Disease Research Centre.

Microbial Genomics –IMB⁴⁸ is involved in the study of microbial ecology and evolution having helped to pioneer the use of culture-independent molecular methods to characterise microbial communities including marker gene and shotgun (metagenomic) approaches, with emphasis on gastrointestinal and skin diseases.

Key UQ personnel, with expertise in the above areas, are listed at **Attachment A**.

Research infrastructure

Recently UQ created the UQ Genome Innovation Hub (GIH), to support UQ researchers and their collaborators in genome research⁴⁹. The GIH works with researchers to develop innovative solutions for complex problems: integrating whole genome analyses (assembly of genomes and transcriptomes), population genetics, single cell genomics (gene expression, epigenetics, gene editing) and functional genomics (gene editing to create cell, organoids and whole animal model systems).

Research interests and capabilities are in medicine, food security and the environment, and cross disciplinary activities.

This is all possible by GIH and other core UQ facilities working together to provide capabilities for data generation and data analysis. At UQ, next-generation sequencing (NGS) facilities at IMB, the Australian Centre for Ecogenomics (ACE) and the Integrated genomics facility – a UQ/ Australian Genome Research Facility (AGRF) collaboration – provide assays for genomes for a range of plant, animal and microbe species.

UQ has facilities to both generate and analyse single cell genomics (IMB, QBI) including cell biology and cell sorting. This work is complemented by facilities for creation of cell lines (stem cells, induced pluripotent stem cells or iPSC) and organoids, at the AIBN.

New traits are of wide interest, facilitated by bioimaging at the Centre of Advanced Imaging (CAI) and the Herston Imaging Research Facility (HIRF) as are new therapies, such as theranostics combining bioimaging and target drugs.

⁴⁸ In Microbial Genomics, the key personnel is Professor Phil Hugenholtz at IMB.

⁴⁹ <https://www.uq.edu.au/news/article/2018/05/uq-leads-way-health-new-genome-innovation-hub>

For functional studies on specific genes and regulatory elements, or specific effects of genome variants, the creation of cell, organoids and whole animal models is important. This is facilitated by the Queensland Facility for Advanced Genome Editing (QFAGE), working at single cells and whole animals (such as mice, zebra fish) levels.

Further work in the development of new tools for diagnostics and prediction depend on the development of new IT and technologies, under development at AIBN and other units.

New therapies depend on use of such models as described above but also new molecules using protein expression facilities at the AIBN, biomarkers (metabolomics, mass spectrometers) and clinical trials using infrastructure supported at TetraQ, IMB, and UniQuest.

Skills and resources in bioinformatics are critical. UQ infrastructure includes access to extensive storage and high performance computing, including at UQ's Research Computing Centre (RCC) and the Queensland Cyber Infrastructure Foundation (QCIF) and skills through the Queensland Facility of Advanced Bioinformatics (QFAB).

A general list of UQ research facilities can be found at <https://research.uq.edu.au/about/facilities-initiatives>.

Collaborations

There are many examples of research collaborations at UQ that have led to commercial products and translation. Of relevance here is the goal of the Queensland Genomics Health Alliance (QGHA) and its close partnership with the Australian Genomics Health Alliance (AGHA) to implement genomics into clinical practice. The following are two examples of work from these genome alliances used to illustrate the translation of genomics into clinical practice.

- **Early Detection of Melanoma:** Professor Peter Soyer is co-leader of the Australian Skin and Skin Cancer (ASSC) Research Centre (UQ and QIMR Berghofer). This centre is developing combined approaches, including 3D total body imaging, a telehealth network and whole exome panels for the implementation of screening and surveillance of high-risk individuals. This work is the result of collaborations between UQ, QIMR and many hospitals funded

through schemes from the Medical Research Future Fund, NHMRC, QGHA with many additional projects funded through other competitive, industry and philanthropic funding sources. In total, he has achieved over \$24 million in research funding for UQ.

- **Rare Genetic Forms of Kidney Disease:** Associate Professor Andrew Mallett is national director of KidGen and co-lead of the Queensland Renal Genetics Service based at the Royal Brisbane and Women's Hospital (RBWH), the first comprehensive renal genetics clinical service in Australia, opened in 2013. It provides multidisciplinary and sub-specialty care in diagnostics and management for Queensland patients with rare and genetic forms of kidney disease. As part of AGHA, Andrew Mallett leads the renal genetics rare disease flagship, providing early diagnosis for patients possibly needing kidney transplants. This is a collaboration of five states (QLD, NSW, VIC, SA and WA), 13 hospitals and six Universities and Research Institutes.

The University of Queensland has many national and international collaborations in genomics (in general) and precision medicine (in particular).

- For the study of neurological and reproductive conditions, IMB/QBI are members of several consortia, including the Psychiatric Genomics Consortium, Australia Systems Genomics Consortium, Centre for Cognitive Ageing and Cognitive Epidemiology (University of Edinburgh, UK), and Australian Coalition for Endometriosis.
- Together with other partners in Queensland – UQ, QIMR, Princess Alexandra Hospital (PAH) and RBWH – and the rest of Australia, UQ and Queensland Health are committed to the implementation and translation of clinical genomics, through a number of genome health alliances, QGHA, AGHA and the Melbourne Genomics Health Alliance, and internationally through the Global Alliance for Genomics and Health (GA4GH).
- The creation of biomedical models of normal and diseased human

states has been facilitated through various consortia, including Stem cells Australia and the Australian Functional Genomics Network.

- UQ also collaborates with many international consortia to access and contribute datasets and novel analytical tools for world-wide use,

(Illumina, PacBio, Oxford Nanopore, Agilent Technologies, Integrative Sciences, Dovetail, Millennium Sciences, 10x Genomics and Neogen) to promote collaborative research with the UQ Genomics Community and collaborators world-wide.

Recently, UQ launched the “UQ Genome Innovation Hub”, a partnership with research institutes at UQ, Faculties and hospitals, with support/collaboration from industry to promote collaborative research with the UQ Genomics Community and collaborators world-wide.

such as the UK Biobank, UK Medical Research Council Health Data Research, and through EMBL-ABR links with Ensembl/EBI and NCBI. The latter is particularly important for assembly and annotation of genomes.

- Recently, UQ launched the “UQ Genome Innovation Hub”, a partnership with research institutes at UQ (IMB, QBI and Queensland Alliance for Agriculture and Food Innovation – QAAFI), Faculties (of Medicine, and of Science) and hospitals (RBWH and PAH), with support/collaboration from industry

For further examples, please refer to websites for:

- Institute for Molecular Bioscience (IMB) – <https://imb.uq.edu.au/>
- Australian Institute for Bioengineering and Nanotechnology (AIBN) – <https://aibn.uq.edu.au/>
- Queensland Brain Institute (QBI) – <https://qbi.uq.edu.au/>
- UQ Faculty of Medicine (FoM) – <https://medicine.uq.edu.au/>
- Queensland Alliance for Agriculture and Food Innovation (QAAFI) – <https://qaafi.uq.edu.au/>

Attachment A:

Go8 key personnel

The Australian National University

Dr Thomas Daniel (Dan) Andrews – Genome Informatics (<https://jcsmr.anu.edu.au/people/academics/dr-thomas-daniel-dan-andrews>)

Professor Emily Banks – Epidemiology and Population Health (<http://rsph.anu.edu.au/people/academics/professor-emily-banks>)

Dr Edward Bertram – Phenomics (<https://jcsmr.anu.edu.au/people/academics/dr-edward-bertram>)

Dr Gaétan Burgio – Genome Editing and Genetics of Host-Pathogen Interactions (<https://jcsmr.anu.edu.au/people/academics/dr-gaétan-burgio>)

Professor Matthew Cook – OHIOH, Personalised Immunology (<https://jcsmr.anu.edu.au/people/academics/professor-matthew-cook>)

Professor Simon Easteal – Indigenous Genomics (<https://jcsmr.anu.edu.au/people/academics/professor-simon-easteal>)

Professor Simon Foote – Director, JCSMR; Genetics and Infectious Diseases (<https://jcsmr.anu.edu.au/people/academics/professor-simon-foote>)

Professor Ross Hannan – Cancer Biology & Therapeutics (<https://jcsmr.anu.edu.au/people/academics/professor-ross-hannan>)

Dr Riccardo Natoli – Clear Vision Research Lab (<https://jcsmr.anu.edu.au/people/professional-staff/dr-riccardo-natoli>)

Professor Thomas Preiss – RNA Biology (<https://jcsmr.anu.edu.au/people/academics/professor-thomas-preiss>)

Professor Eric Stone – Quantitative and Computational Biology (<http://biology.anu.edu.au/people/eric-stone>)

Professor Lyndall Strazdins – Epidemiology and Population Health (<http://rsph.anu.edu.au/people/academics/professor-lyndall-strazdins>)

Professor David Tremethick – Genome Science (<https://jcsmr.anu.edu.au/people/academics/professor-david-tremethick>)

Associate Professor Antonio Tricoli – OHIOH, phenotype monitoring (<https://eng.anu.edu.au/people/antonio-tricoli>)

Professor Carola Vinuesa – Personalised Immunology (<https://jcsmr.anu.edu.au/people/academics/professor-carola-g-vinuesa>)

Professor Catherine Waldby – Social studies of biomedicine and the life sciences (<https://researchers.anu.edu.au/researchers/waldby-c>)

UNSW Sydney

Professor Susan Ramus – School of Women's and Children's Health, Faculty of Medicine (ovarian cancer: genetic susceptibility and identification of prognostic markers)

Professor Merlin Crossley – Deputy Vice Chancellor Education and School of Biotechnology and Biomolecular Sciences, Faculty of Science (inherited blood diseases)

Professor John Pimanda – Head of Department of Pathology and Adult Cancer Program, Prince of Wales Clinical School and School of Medical Sciences, Faculty of Medicine

Professor Emad M El-Omar – Director, Microbiome Research Centre, St George & Sutherland Clinical School, Faculty of Medicine

Associate Professor Fabio Luciani – Kirby Institute for Infection and Immunity and School of Medical Sciences, Faculty of Medicine (immunology)

Dr Irina Voineagu – School of Biotechnology and Biomolecular Sciences, Faculty of Science (autism spectrum diseases)

The University of Adelaide

Professor Timothy Hughes – determinants of response and resistance to therapy in leukaemia, with a particular focus on chronic myeloid leukaemia (CML)

Professor Jozef Gecz – founding Head of the Neurogenetics Research Program (=NEURO)

Emeritus Professor Alastair MacLennan – leads the Australian Collaborative Cerebral Palsy Research Group with Professor Jozef Gecz

Professor Paul Thomas – Director of the South Australian Genome Editing (SAGE) Facility and Head of the Genome Editing Laboratory at the University of Adelaide and South Australian Health and Medical Research Institute (SAHMRI)

Professor Deborah White – lead investigator of the Australian Genomics Health Alliance (AGHA: Australian Genomics) Acute Lymphoblastic Leukaemia (ALL) Flagship, and SA scientific lead for Zero Children's Cancer (ZCC)

Professor Claire Roberts and Professor Gus Dekker – lead the Placental Development Group

Professor Andrew Somogyi and Dr Dan Barrett – pharmacogenomics

The University of Western Australia

Professor Nigel G Laing AO – Group Leader, The Neurogenetic Diseases Group

Professor Alistair Forrest – Group Leader, The Systems Biology and Genomics Lab

Dr Ryan Lister – Group Leader for Characterising and Editing the Epigenome

Professor Eric Moses – GoHAD Group Leader

Professor Wendy Erber – Translational Cancer Pathology Group Leader

Professor David Mackey – Genetic Eye Diseases Group Leader

Associate Professor Joost Lesterhuis – Immuno-oncology Group Leader

Professor Anna Nowak – National Centre for Asbestos Related Diseases Group Leader

Attachment A: Go8 key personnel

The University of Sydney

Professor Ian Alexander Head – Gene Therapy Research Unit, CMRI and CHW

Associate Professor Bruce Bennetts – Head, Sydney Genome Diagnostics, CHW

Professor David Booth – Director of Research, Centre for Immunology and Allergy Research, Westmead Institute for Medical Research (WIMR); Research Coordinator, MD program, Westmead Clinical School, University of Sydney

Associate Professor Sandra Cooper – NHMRC CDF, Paediatrics & Child Health, CHW; Deputy Head, Kids Neuroscience Centre, Kids Research, CHW; Head, Functional Neuromics, CMRI

Professor Tony Cunningham – Executive Director of WIMR and of its Centre for Virus Research; Director, Australian Centre for HIV, Hepatitis and HTLV1 Virology Research; President, AAMRI

Professor Anna deFazio – Sydney West Chair in Translational Cancer Research, University of Sydney; School Academic Leader (Research), Sydney Medical School; Co-Deputy Director, Sydney-West Translational Cancer Research Centre; Laboratory Head, Centre for Cancer Research, WIMR and Department of Gynaecological Oncology, Westmead Hospital

Professor Jacob George – Robert W Storr Chair of Hepatic Medicine, Faculty of Medicine and Health; Director, Storr Liver Centre, WIMR; Head, Department of Gastroenterology & Hepatology, Westmead Hospital and Sydney West Local Health District

Professor Glenda Halliday – NHMRC Senior Principal Research Fellow & Head, ForeFront Research Program on Ageing and Neurodegeneration, Brain and Mind Centre

Professor Vanessa Hayes – Petre Chair of Prostate Cancer Research, Central Clinical School, Faculty of Medicine and Health

Professor Edward C. Holmes – Professor and Laureate Fellow, Faculty of Science and Medicine and Health; Marie Bashir Institute for Infectious Diseases and Biosecurity (MBI)

Professor Jonathan Iredell – Senior Staff Specialist Infectious Diseases and Microbiology, Westmead Hospital; Director of Centre for Infectious Diseases & Microbiology (CIDM), WIMR; NHMRC Senior Practitioner Research Fellow and Professor of Medicine and Microbiology (conj.), University of Sydney

Professor David James – Leonard P Ullmann Chair in Metabolic Systems Biology, Metabolic Cybernetics Lab, The Charles Perkins Centre, School of Life and Environmental Sciences and Sydney Medical School, University of Sydney

Professor Robyn Jamieson – Head of the Eye Genetics Research Unit, CMRI (Laboratory component), and CHW & Save Sight Institute

Professor Christopher Liddle – Professor of Clinical Pharmacology and Hepatology, University of Sydney, Westmead Hospital; Principal Investigator, Storr Liver Centre, WIMR; Adjunct Professor, Salk Institute for Biological Studies, La Jolla CA, USA

Dr Leszek Lisowski – Head, Vector and Genome Engineering Facility, and Translational Vectorology Group, CMRI

Professor Georgina Long – Chair of Melanoma Medical Oncology & Translational Research Northern Clinical School; Co-Medical Director of Melanoma Institute Australia

Professor Graham Mann – Professor of Medicine, Westmead Clinical School; Co-Director, Centre for Cancer Research, WIMR; Chair, University of Sydney Cancer Research Network

Professor Wieland Meyer – Professor of Molecular Medical Mycology Medicine, Westmead Clinical School, Faculty of Medicine and Health, MBI; Head of the Molecular Mycology Laboratory, Centre for Infectious diseases and Microbiology, WIMR

Professor Michael Murray – Professor of Pharmacology, Faculty of Medicine and Health

Associate Professor Greg Neely Associate Professor, School of Life and Environmental Sciences, Faculty of Science

Professor Roger Reddel – Sir Lorimer Dods Professor, Faculty of Medicine and Health; Director, CMRI; Head, Cancer Research Unit, CMRI

Professor Christopher Semsarian – NHMRC Practitioner Fellow; Program Head and Faculty, Molecular Cardiology, Centenary Institute

Professor Vitali Sintchenko – Professor, Sydney Medical School; Director, Centre for Infectious Diseases and Microbiology-Public Health, Westmead Hospital and NSW Health Pathology

Professor Tania Sorrell – Director, Marie Bashir Institute for Infectious Diseases and Biosecurity; Professor, Clinical Infectious Diseases; Deputy Dean (Clinical), Faculty of Medicine and Health; Group Leader, Centre for Infectious Diseases and Microbiology, WIMR; Centre for Infectious Diseases & Microbiology (Public health)

Professor Carolyn Sue – Director of Neurogenetics & Neurologist & Head of Neuroscience Research, Kolling Institute

Professor Graeme Stewart – Director of the Centre for Immunology and Allergy Research, WIMR; Director, Clinical Immunology & Allergy, Westmead Hospital; Head, Ambulatory Services, Westmead Hospital

Professor Robyn Ward – Executive Dean, Faculty of Medicine and Health

The University of Melbourne

Professor Sean Grimmond – Precision Oncology Leader

Professor Kathryn North, Associate Professor Kathryn Holt – Microbial/pathogen genomics Leaders

Professor John Hopper – Genetic epidemiology Leader

Professor Ingrid Scheffer and Professor Sam Berkovic – Genetic diagnostics of epilepsy Leaders

Professor David Balding and Associate Professor Stephen Leslie – Statistical Genomics Leaders

Attachment A: Go8 key personnel

Professor James McCluskey
– Immunogenetics Leader

Professor Ingrid Winship
– Clinical Genetics Leader

Professor Darren Kelly
– founder Certa Therapeutics

Professor Alastair Stewart – ARC Training
Centre for Personalised Therapeutic
Technologies

Monash University

Professor Melissa Southey – Chair of
Precision Medicine at the School of Clinical
Sciences at Monash Health

Professor Mark Shackleton Director of
Oncology at Alfred Health and Professor Gail
Risbridger, Director of Research at the Monash
Partners Comprehensive Cancer Consortium
– oversee Precision Oncology program

Professor Tony Purcell –
Immunology and Infectious Diseases

Professor Andrew Wei, Professor Andrew
Perkins and Professor David Curtis –
Clinical haematologists

Associate Professor Sefi Rosenbluh –
Functional cancer genomics and the role
of cellular signalling in cancer

Dr Paul Lacaze – The Public Health
Genomics Program

Professor Patrick Kwan and Professor Terry
O'Brien – leading researchers in epilepsy

Professor Helmut Butzkueven
– Multiple Sclerosis

Professor Mark Bellgrove – genetic factors
behind conditions such as attention deficit
hyperactivity disorder (ADHD)

Professor Sam El-Osta – diabetes
and cardiovascular diseases

The University of Queensland

Professor David W. Burt – Director
UQ Genomics, Office of Deputy Vice
Chancellor Research

Professor Peter Visscher, Professor Naomi
Wray, Professor Jian Yang and Professor
Grant Montgomery (IMB), Professor David
Evans (UQ Diamantina Institute) – Statistical
Genomics of Complex Traits

Dr Nathan Palpant, Associate Professor
Ben Hogan and Professor Peter Koopman
(IMB) and Professor Ernst Wolvetang (AIBN)
– Functional Genomics

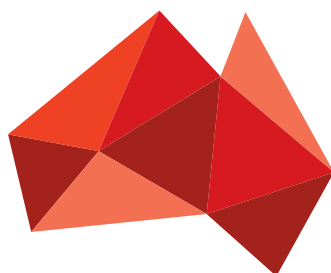
Dr Sam Lukowski and Dr Quan Nguyen
(IMB) – Single Cell Genomics

Professor David Paterson, Professor
Mark Walker, Professor Mark Schembri
and Associate Professor Scott Beatson
(Australian Infectious Disease Research
Centre) – Infectious Diseases Genomics

Professor Phil Hugenholtz
– Microbial Genomics

Professor Peter Soyer – co-leader the
Australian Skin and Skin Cancer (ASSC)
Research Centre (UQ and QIMR Berghofer)

Associate Professor Andrew Mallett –
National director of KidGen and co-lead of
the Queensland Renal Genetics Service based
at the Royal Brisbane and Women's Hospital



GROUP OF EIGHT AUSTRALIA

MEMBERS



THE UNIVERSITY OF
**WESTERN
AUSTRALIA**



MONASH
University



Australian
National
University



THE UNIVERSITY
of **ADELAIDE**



THE UNIVERSITY OF
MELBOURNE



UNSW
SYDNEY



THE UNIVERSITY
OF QUEENSLAND
AUSTRALIA



THE UNIVERSITY OF
SYDNEY

go8.edu.au

