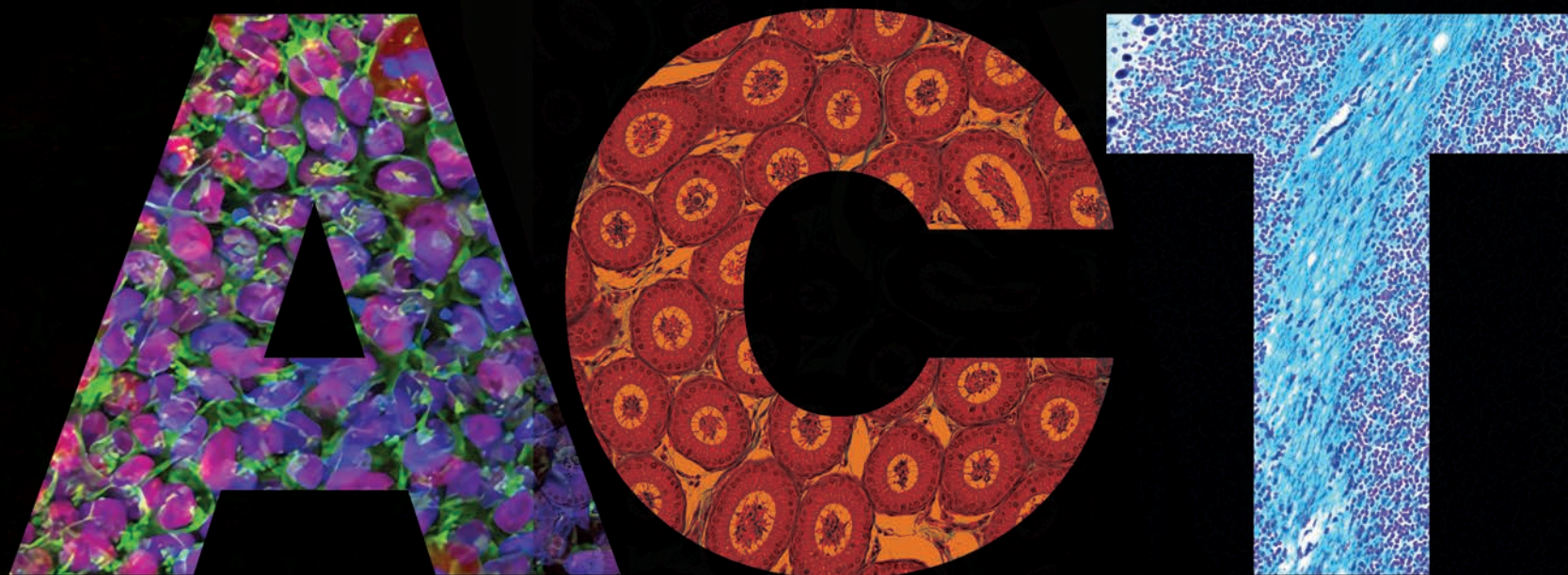




2023



Acknowledgement

Phenomics Australia acknowledges First Nations people as the Traditional Custodians of the lands on which this work was imagined and realised. We pay our respects to their Elders past and present.

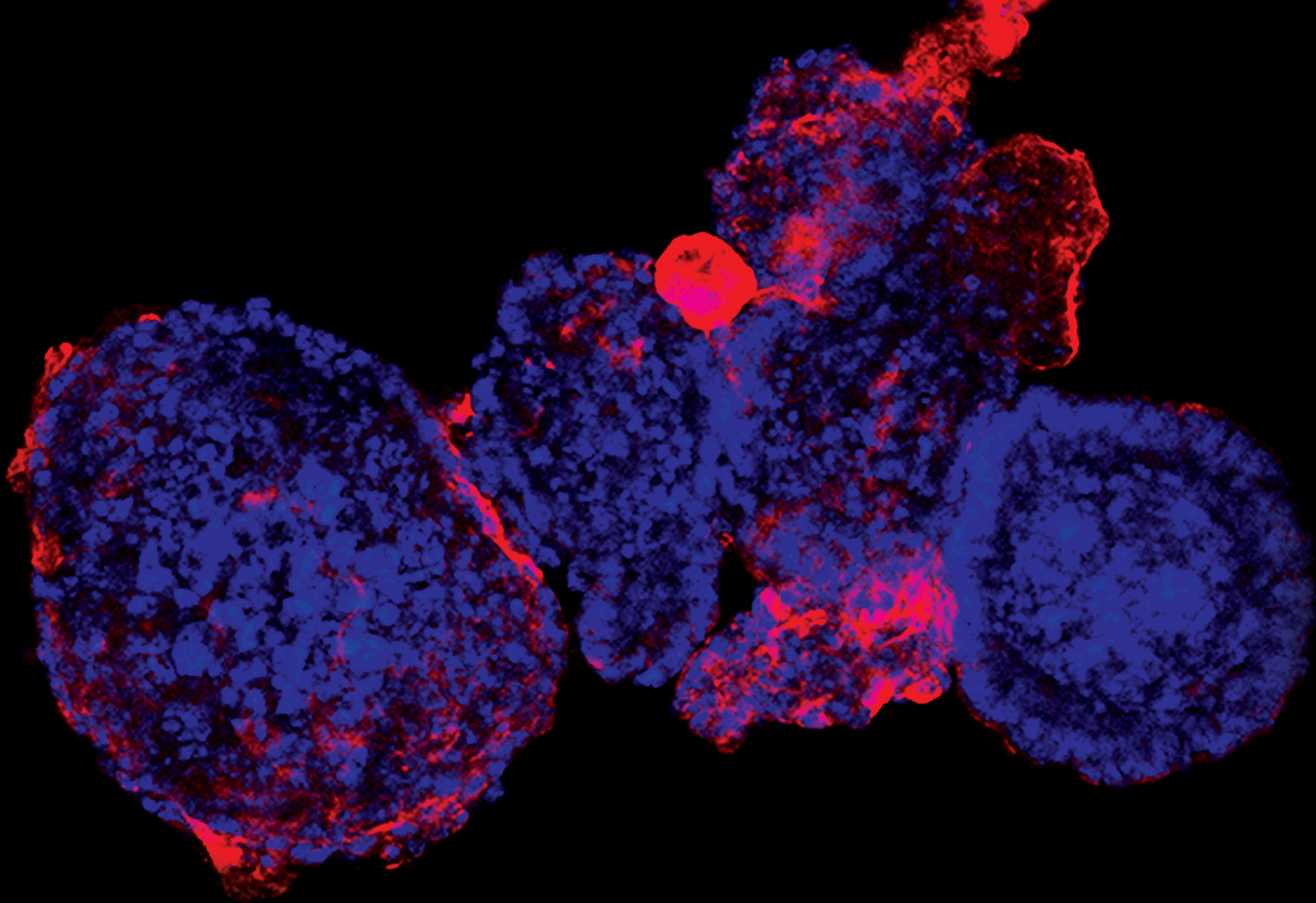
Thank you to our Phenomics Australia team members and host institutions for their contributions to this publication.

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Image credit: Confocal image of intestinal organoid by Jie Tang.

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Editorial: Dr Marina Trigueros



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Chair Report

As the Chair of the Phenomics Australia Board, I am delighted to share with you the remarkable achievements of our team and researchers, reflecting our commitment to excellence and innovation in research infrastructure provision.

Foremost, we are very proud of the successful award of \$42 million in additional Commonwealth Government funding to preserve the ongoing operation of Phenomics Australia into the future. This significant financial injection not only strengthens our foundations but also positions us as a formidable force in driving transformative initiatives to roadmap national research infrastructure in Australia. In particular, this new funding will allow us to spearhead the rollout of new national services in synthetic biology. This move reflects our proactive approach to stay at the forefront of technological evolution, positioning us as leaders in research and development, and considering disciplines beyond health that application of our expertise can be of significant value. The synthetic biology initiative is a testament to our commitment to cutting-edge solutions that address the evolving needs of our stakeholders.

Our rollout of new funding mechanisms to support service vouchers has proven to be instrumental in enabling early-to-mid-career researchers to develop their work at the crucial proof-of-concept stage, particularly where national research council support may not be forthcoming, and I congratulate all the successful voucher recipients for their hard work.

In tandem with these developments, we have witnessed a substantial surge in usage of Phenomics Australia services, signifying

the growing influence and impact we have on the community. This not only validates the relevance of what we offer but also serves as a testament to the trust and confidence our users place in us. I am pleased to report that this pursuit of excellence has not gone unnoticed with this expanding user base, with extremely high user satisfaction a direct result of our team's dedication and the strategic vision that guides what we do. This positive feedback underscores the impact and relevance of our services in addressing critical challenges faced by our community.

I commend and recognise our strongest assets – our dedicated team of over 85 specialist technical staff, who have been instrumental in delivering nearly 40 specialized services across 20 locations. Their expertise forms the backbone of our success, ensuring that we provide comprehensive and tailored solutions that resonate with the diverse needs of our stakeholders.

I look forward to continued progress and future accomplishments as we navigate the exciting opportunities that lie ahead.



Dr. Liz Jazwinska
Board Chair,
Phenomics Australia

CEO Report

I am very pleased to present to you the Phenomics Australia 2023 Impact Report which highlights some of the investments, activities, and scientific advances that have been enabled through the NCRIS program. These are a testament to the value of Phenomics Australia's world-class services and expertise and celebrate the dedication and skilled contributions of each member of the Phenomics Australia network of technicians, scientists, advisors, governors, and executives that are hosted by our 12 partner universities and medical research institutes.

The award of \$42 million in additional Commonwealth Government funding for 2023-2028 reflects our proactive approach to staying at the forefront of technological evolution and reaffirms our position as leaders in research and innovation. Beyond the health sector, our expertise is recognised for its significant value, showcasing our commitment to cutting-edge solutions.

As we welcome the government's support for our strategy, providing continuity across the national research infrastructure landscape, I am excited about the prospects that lie ahead. We look forward to continuing our collaborative efforts with all stakeholders to progress the important issues outlined in the Research Infrastructure Investment Plan.

A major focus for 2022-23 was to complete the establishment of Australia's first In Vitro Genome Engineering and Disease Modelling Service. We have also been working to establish Australia's first national Drosophila Transgenic Facility. Together, these represent the necessary and valuable expansion of the range of model systems that are available to Australian researchers at the

required quality and scale.

In September, we had the pleasure of enjoying our first face-to-face strategic leadership retreat in three years. This retreat provided a valuable opportunity for our leadership team to come together, exchange ideas, and chart the course for the future. It reinforced the strength of our team and highlighted the collective determination to achieve our goals.

In conclusion, I am very pleased to present the Phenomics Australia Impact Report to you and am excited about what can be achieved in the coming year. Our journey is marked by excellence, collaboration, and a shared commitment to advancing research and innovation. I am confident that with our outstanding team, we will continue to make meaningful contributions to the scientific community and beyond.



Prof. Michael Dobbie
CEO,
Phenomics Australia



Top left: Phenomics Australia Executive Team at the Australian National University

Top right: Phenomics Australia node the Victorian Centre for Functional Genomics (VCFG) team at Peter MacCallum Cancer Centre

Bottom left: Phenomics Australia node the South Australian Genome Editing (SAGE) facility team at South Australian Health and Medical Research Institute

Centre: Phenomics Australia Strategic Partnerships Advisor at AusBiotech (with a large quokka)

About Phenomics Australia

Phenomics Australia provides academia and industry with genome engineered cell, tissue and animal pre-clinical models necessary to study disease, underpinning what is widely known as Precision Medicine. Our experts help researchers study their models and manage their research projects to accelerate discovery and understanding of what genes do, find genetic causes of diseases, develop new therapeutics and create better health treatments.

Our technologies and expertise are also increasingly applicable beyond the area of health, and we regularly provide services for projects in defence, modern manufacturing, environment, biosecurity, and agriculture.

We offer a broad portfolio of cell- and animal-based disease model systems, genome engineering, biochemical and molecular screening, and biobanking services, user training, and technical expertise through a national alliance of capabilities and partner institutions.

Vision

Better health through the discovery of gene function.

Mission

Providing world-class infrastructure and expertise, collaborating for research excellence, and advocating and partnering for impact, Phenomics Australia will drive the discovery of the molecular basis of disease to benefit the health of all Australians.

Aspirations

Building and delivering world-class infrastructure and expertise

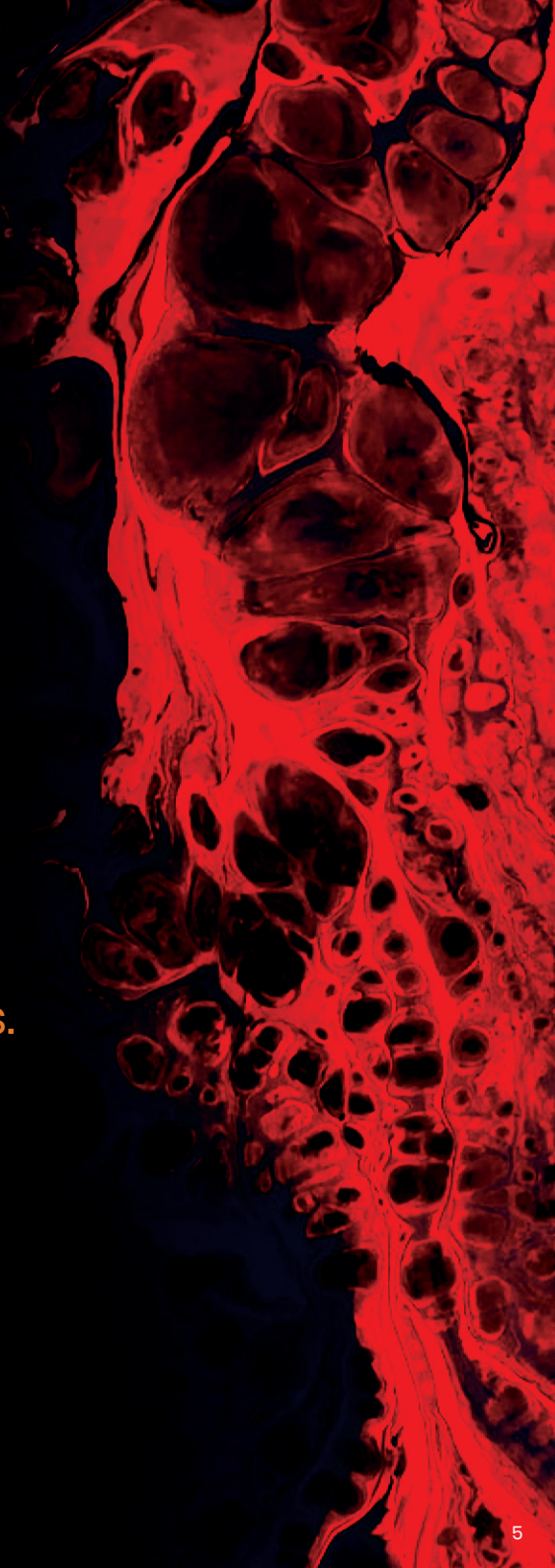
We will deliver cutting-edge, versatile, and sustainable phenomics infrastructure to accelerate discovery and translation.

Collaborating for research excellence

We will partner to deliver innovative research outcomes; to advance our understanding of health and disease and to inform next-generation healthcare and clinical practice.

Advocating and partnering for impact

We will reaffirm Phenomics Australia as a thought-leader and influencer within the research and policy landscape.



Our Partners



Australian National University



Peter Mac
Peter MacCallum Cancer Centre
Victoria Australia



SAHMRI
South Australian Health & Medical Research Institute



MONASH University



murdoch children's research institute



THE UNIVERSITY of ADELAIDE



Perkins
HARRY PERKINS INSTITUTE OF MEDICAL RESEARCH



WEHI
brighter together



THE UNIVERSITY OF QUEENSLAND AUSTRALIA



Victor Chang
Cardiac Research Institute



VICTOR CHANG CARDIAC RESEARCH INSTITUTE INNOVATION CENTRE



THE UNIVERSITY OF MELBOURNE



NCRIS
National Research Infrastructure for Australia
An Australian Government Initiative


Phenomics Australia is supported by the Australian Government through the National Collaborative Research Infrastructure Strategy, NCRIS. NCRIS supports Australia's research capability by investing in research infrastructure and making it accessible to researchers across the nation.

Our Service Locations




By the Numbers


22 –
23

 **1,409**
Users

 **20**
Locations


 **\$43m**
New Government
funding announced

 **38+**
Research
services

 **20**
Different fields
of research

 **85+**
Technical
experts

 **\$23m**
Asset
valuation

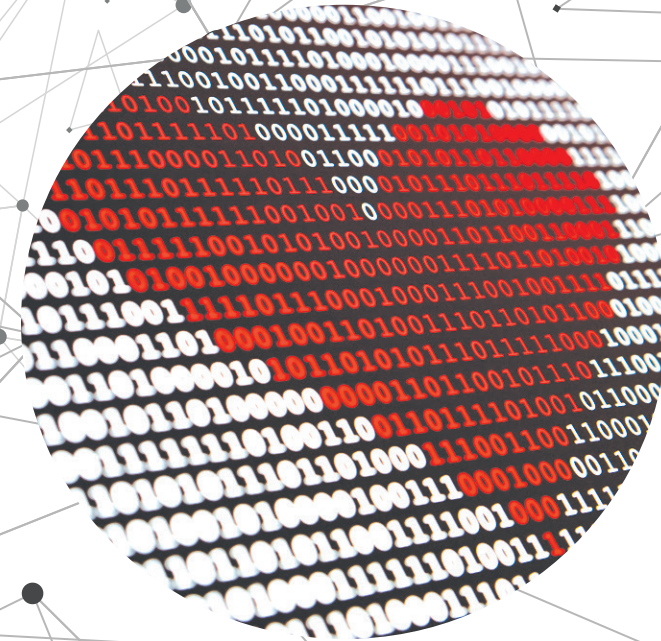
 **\$72m**
Research grants
supported

96% Of users report
Phenomics
Australia
improved their
research quality

96% Of users would
recommend
our facilities



Top: ANU Centre for Therapeutic Discovery



Our Collaborations



Infrastructure Partnerships

NCRIS Health Group

Health and medical researchers investigate solutions to complex problems that are often not easily solved by a single discipline. The NCRIS Health Group is enabling them collaboratively throughout the innovation and translational process.

TIA

Phenomics Australia and Therapeutic Innovation Australia (TIA) collectively offer academic researchers and SMEs access to a diverse range of Australian translational medical research capabilities, through the Pipeline Accelerator scheme

Biomedical Data Asset

Linking two of Australia's most important biomedical datasets in a highly curated user-friendly and accessible environment: the Biomedical Data Asset has been developed with ARDC, BPA, Monash and ANU.



Research and Academic

AFGN

Advancing Functional Genomics with the Australian Functional Genomics Network (AFGN) as infrastructure provider of choice.

Rare Care Centre

Addressing the challenges of Persons living with a rare disease and their Families, with The Rare Care Centre at Perth Children's Hospital in Western Australia.



Industry Alliances

CSIRO

Delivery of the CSIRO Futures report setting out Australia's opportunities for medical product development through the use of non-animal models.

OzgeneARC

Supporting continuity of national laboratory animal supply with OzgeneARC.



International Partnerships

IMPC and Infrafrontier

Advancing biomedical science through in vivo disease models and databases with the International Mouse Phenotyping Consortium (IMPC) and Infrafrontier.

Austrade

Promoting biotechnology exchange between Korea and Australia.

Australian and European Health Research Infrastructures

Strengthening collaboration between European and Australian research infrastructures with NCRIS members.



Community Engagements

NCRIS Comms

Working with the NCRIS community to develop outreach strategies and awareness of our national research infrastructure.

ANU and CPAS

Phenomics Australia hosted a Science Communication Internship in collaboration with the Australian National Centre for the Public Awareness of Science (CPAS) at the Australian National University (ANU).

EMBL A

Inspiring PhD students with the latest ideas and advances in molecular biology and the life sciences with EMBL Australia.



Government and Policy

The National Research Infrastructure Advisory Group

Advise on priorities, trends and investment opportunities to ensure that researchers have access to cutting-edge infrastructure.

Department of Education

Strategy development and investment planning to enable the 2021 NRI Roadmap.

Australian Academy of Science

Scoping work is in response to the 2021 NRI Roadmap to develop a response to infrastructure needs in collections and biobanking.



Top left: European and Australian research infrastructure teams.

Top right: BioKorea 2023 conference.

Bottom left: ANU Centre for Therapeutic Discovery.

Supported Programs

National Health and Medical Research Council (NHMRC)
 Australian Research Council (ARC)
 Enabling Openness in Australia Stem Cell Research (EOAR)
 Medical Research Future Fund (MRFF)
 Ovarian Cancer Research Foundation
 Passe & Williams Foundation
 Robinson Research Institute (RRI)
 Tour de Cure
 The Hospital Research Foundation (THRF)
 Juvenile Diabetes Research Foundation (JDRF)
 ACT Cancer Council
 Bruce and Jenny Pryor
 US Department of Defense
 Bill and Melinda Gates Foundation
 reNEW
 Carina Biotech
 The Michael J. Fox Foundation for Parkinson's Research
 Canberra Health Services
 Anaxis Pharma
 Genentech
 CSL Limited

2022



Phenomics Australia welcomed Dr Twishi Gulati as the new National *In Vitro* Service Coordinator.



Phenomics Australia signed the NCRIS Health Group MoU. National research infrastructure working together to improve the health of Australians.



Monash BDI Organoid Program Symposium in Emerging Technologies in Organoid Research was held at Monash University, Melbourne with over 200 participants.

New website was launched for Phenomics Australia's SAGE node.

Phenomics Australia supported the development of the Research Infrastructure Workforce Position Paper alongside other NCRIS members.

Phenomics Australia Board member, Prof Ute Roessner elected as 2022 Fellow of the Australian Academy of Science.

Phenomics Australia user, Prof Si Ming Man, wins Prime Minister's Science prize.

Phenomics Australia Strategy workshop took place in Melbourne.



2023

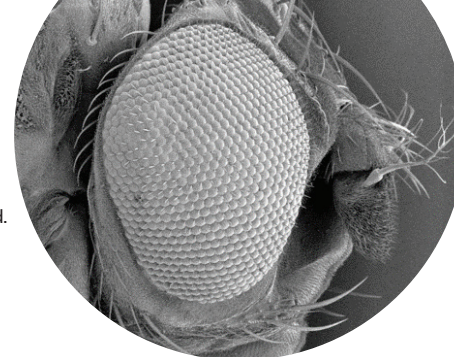


Phenomics Australia's new merchandise arrived.

The newest Phenomics Australia *In Vivo* node, The Australian Drosophila Transgenic Facility launched.

Phenomics Australia joined forces with the Australian Functional Genomics Network (AFGN) for an AFGN-Funding opportunity: FxGx platforms to assess multiple VUS in priority genes.

Phenomics Australia hosted the NRI Advisory Group alongside other NCRIS members at the Australian National University about the Synthetic Biology Revolution.



Phenomics Australia joined forces with the ISSCR to launch the Standards for Human Stem Cell Use in Research.

Phenomics Australia's scientific leader, Prof. Alice Pébay, is awarded an Order of Australia (AM).

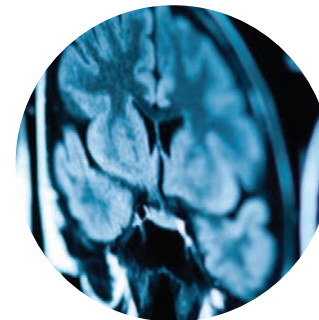
Phenomics Australia supported research gained international media coverage with the news about Kathleen Folbigg's release from prison.

Phenomics Australia IMPACT Voucher is presented at the Organoids Are Us Symposium in the context of empowering scientific breakthroughs supporting access to National Research Infrastructure.

Phenomics Australia joined forces with EMBL Australia to organise the EMBL Australia PhD Course.

The NCRIS Health Group underpins world-first brain cancer collaboration.

Phenomics Australia launched new services flyers.



MAY

Phenomics Australia was at the NCRIS community gathering at ANU – a new initiative to share our work and achievements with the ANU NCRIS community.

Phenomics Australia and other NCRIS members worked with the Australian Academy of Science to roll out The Ukraine-Australia Research Fund.

"Five questions with" interview series presented Dr Susan Pond Phenomics Australia Board member.

Phenomics Australia CEO, Prof. Michael Dobbie, appointed Professor in Practice, National Research Infrastructure.

Phenomics Australia attended the BioKorea 2023 conference forging valuable connections for the NCRIS Health Group.



MAR

"Five questions with" interview series presented Dr Michael Poidinger Phenomics Australia Board member and Data Integrity Committee Chair.

Phenomics Australia celebrated International Women's Day 2023.

Members of Phenomics Australia participated in the 2023 Science and Technology Australia premier event, Science Meets Parliament.

FEB

Phenomics Australia celebrated the International Day of Women and Girls in Science 2023.

Phenomics Australia joined forces with TIA supporting the Pipeline Accelerator Voucher scheme to expand the discovery and translation of medical research.

Phenomics Australia spoke at the Science Communicators Conference in Canberra to discuss what is needed for Communicating Research Infrastructure in Australia.

JAN



DEC

Phenomics Australia *In Vitro* node at VCCRI launched a special end-of-the-year offer to generate induced pluripotent stem cell lines.

Phenomics Australia users were awarded NHMRC grants to help advance understanding of a wide range of health and medical issues faced by Australians.

Phenomics Australia nodes joined forces to exchange best practices and train each other on the Ethical Considerations of Working with Human Biospecimens.

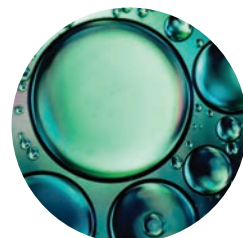


NOV

OCT

Phenomics Australia, EOAR, NHMRC and MRFF joined forces to develop seminars for Enabling Openness in Australian Stem Cell Research.

Phenomics Australia was at the AusBiotech 2022 Conference in Perth with over 1000 participants in attendance.



AUG

Phenomics Australia participates at the National Science Week.

Phenomics Australia hosted a Precision Medicine and Beyond facility tour at APF and ACTD about finding synergies between Research Infrastructures

"Five questions with" interview series published presenting Dr Carol Wicking, Phenomics Australia Science Advisor.



JUL

2023



Phenomics Australia and TIA partnership supported advances in therapeutic development research.

JUL

AUG

Phenomics Australia celebrated its Phenomenal People at National Science Week.

Phenomics Australia and TIA teamed up once more, with voucher support for translational research.

Phenomics Australia sponsored and co-steered the development of a CSIRO Futures report on national opportunities around non-animal modelling.



As an active member of the IMPC, Phenomics Australia invited users to attend the 'Home Cage Monitoring in Behavioral Phenotyping: Opportunities and Feasibility' Workshop.

Phenomics Australia's node the Monash BDI Organoid Program hosted a method-focused symposium on 'Emerging Technologies in Organoid Research'.

Phenomics Australia Strategic Retreat took place.

Phenomics Australia hosted a visit from the Department of Education tour state-of-the-art facilities.

SEP

Phenomics Australia advancing functional genomics through the AFGN's \$1.13M initiative.

"Five questions with" interview presented Prof Glenn Withers Phenomics Australia Board Member and Partner Advisory Committee Chair.

Phenomics Australia Node Director Professor Kaylene Simpson received the Outstanding Leadership by Example Award from the VCCC Alliance.

The NCRIS Health Group visited Europe to strengthen collaboration between European and Australian research infrastructures.

OCT

Phenomics Australia users elected Fellows of the Australian Academy of Science.

Phenomics Australia awarded over \$42m under the Research Infrastructure Investment Plan funding.

Phenomics Australia sponsored the inaugural CRISPR Down Under Symposium.



2023 – Future

Phenomics Australia Scientific Leader Dr Louise Winteringham presented a NSW Health Statewide Biobank seminar.

We explored the heart of Phenomics Australia: A Journey Across Nodes.

NOV

Phenomics Australia and TIA jointly exhibited at ASSCR 2023 'Navigating New Frontiers in Stem Cell Research'.

Phenomics Australia datasets at the centre of the Data Sprint on Imaging Transcriptomics.

Phenomics Australia exhibited at the 2023 ANU Research Infrastructure Expo.



DEC

Phenomics Australia Scientific Leaders shared their knowledge at the 2023 EMBL Australia PhD Course.

FUTURE

Rollout of the Phenomics Australia FAIR Data Strategy and supporting data infrastructure.

Broaden Phenomics Australia's portfolio of national disease modelling capabilities to serve all needs.

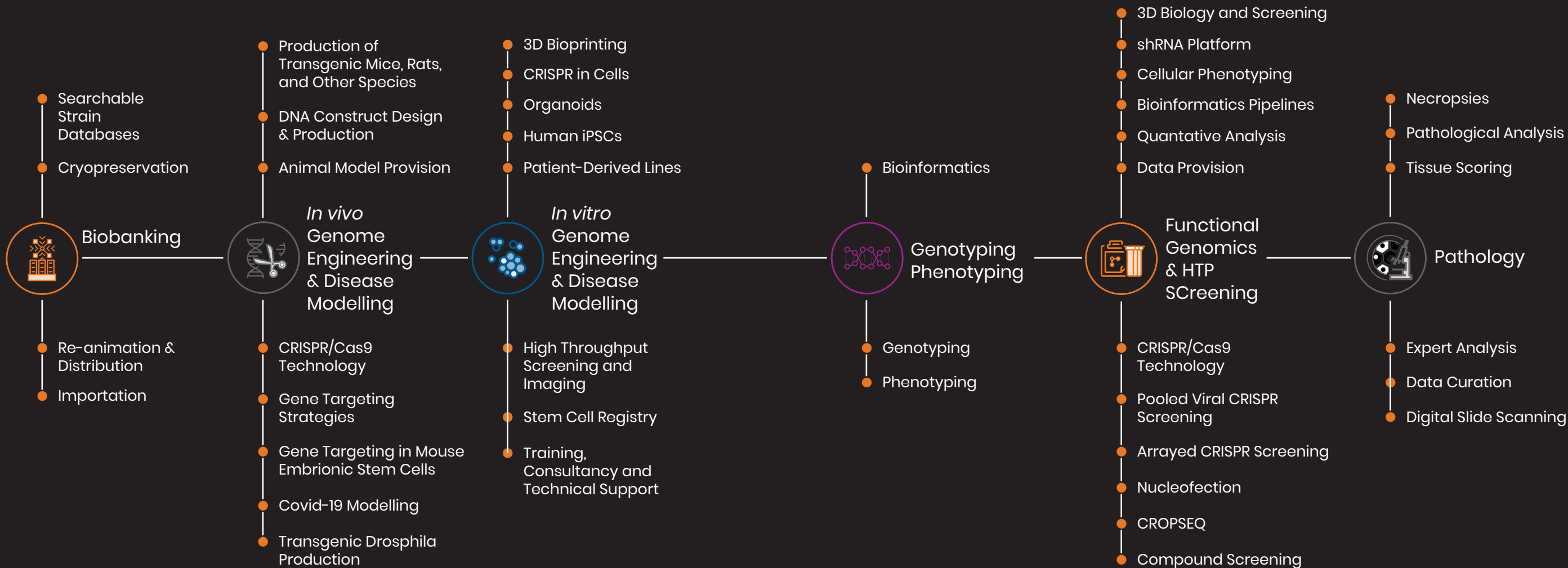
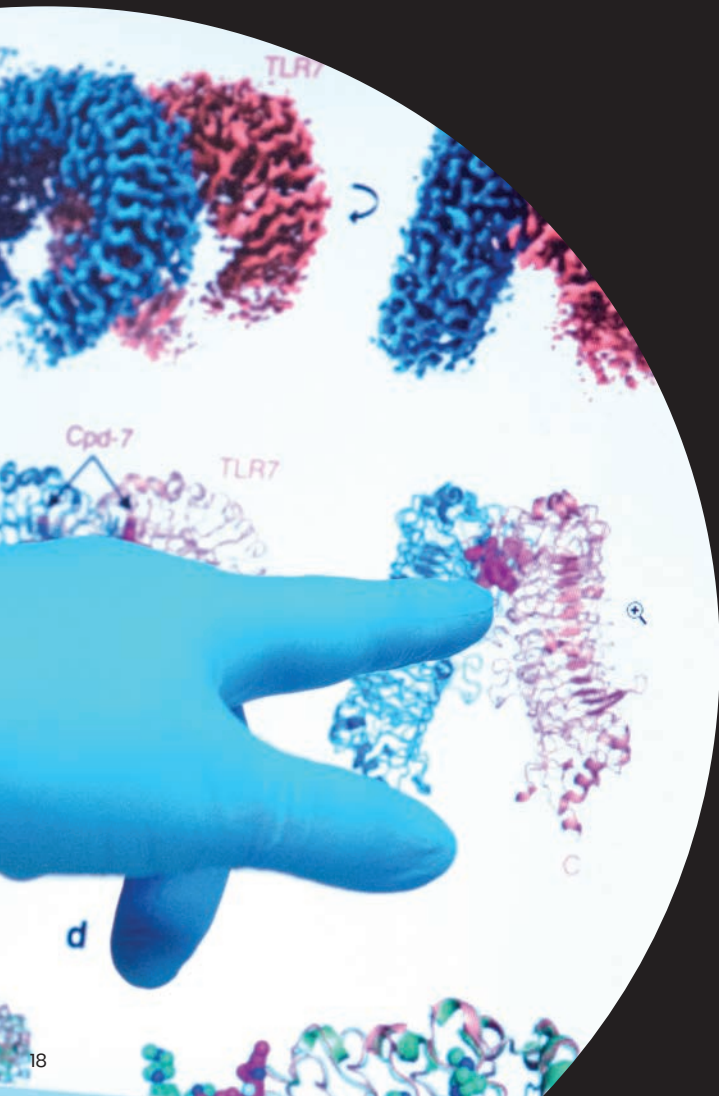
Deploy Phenomics Australia skills and technologies to further enable impact beyond health.

Investment in national research infrastructure supporting Synthetic Biology.

Enable the NRI Roadmap through step change in national infrastructure supporting Digital Research, Research Translation, and Collections.



Our Services



Dr Martin Pal at MAGEC laboratory.

Our Impact

In vitro Genome Engineering and Disease Modelling Service

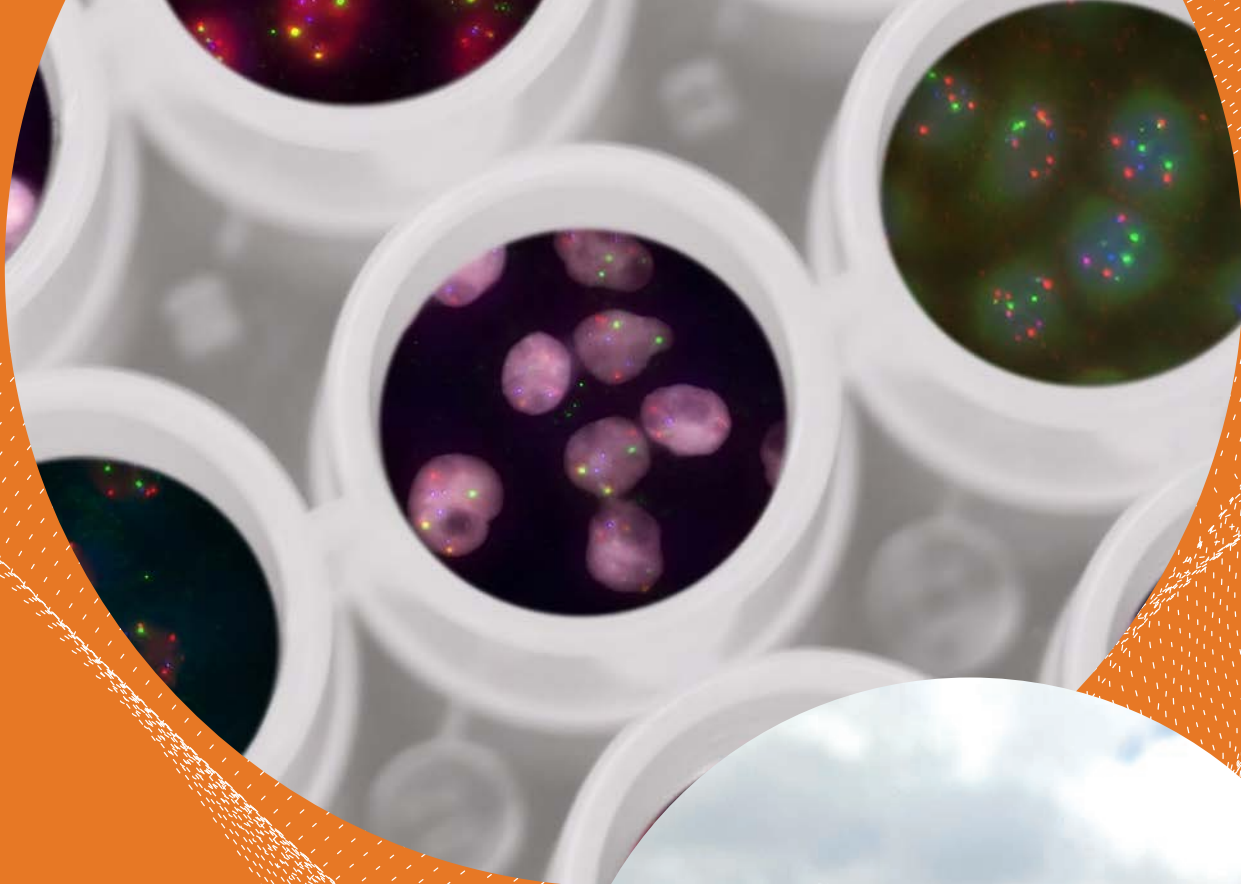
Our new *in vitro* Genome Engineering and Disease Modelling Service will enable Phenomics Australia to offer a more comprehensive and enriched portfolio of sophisticated non-animal modelling technologies.

Through a collaborative consortium of new laboratories and facilities across eight institutions and five states and territories, this service expansion is boosting Australia's capability and capacity to understand the functional consequences of DNA sequence variation in the human genome for health and disease. The synergistic network of the service providers will deliver adaptable and scalable disease-modelling platforms for improved diagnoses, Precision Medicine for genetic disorders, and therapeutic development by both academia and the biopharmaceutical industry

The Australian Drosophila Transgenic Facility

The Australian Drosophila Transgenic Facility is a unique facility for generating transgenic strains of the fruit fly *Drosophila melanogaster*. It utilises the revolutionary CRISPR/Cas9 genome editing technologies complemented with well-established transgenesis methods.

Drosophila is an outstanding model organism for molecular genetic research, and Drosophila research has provided many crucial fundamental biological insights. The striking conservation between fly and human genes and their mechanisms of action has also meant that the study of this organism has provided insights into multiple aspects of human biology.



Biobanking



The Australian Phenome Bank

Phenomics Australia's Biobanking service, run by the Australian Phenome Bank, provides a database and cryobank of mouse sperm and embryos essential for discovering and distributing disease models in Australia, and aiding the advancement of personalised medicine.

The challenge:

Ensuring the ongoing availability of mouse strains preserving the investment made in creating and characterising them and creating a global resource of enormous value.

The solution:

Centralised repositories are essential if the valuable mouse strains and models that have been developed are to be securely preserved and fully exploited.

The future:

Complementing the Australian Phenome Bank with a searchable Australian Stem Cell Registry.

Disease detectives: a new frontier in genetic mutations

New research published in Science Immunology conducted by an international consortium of researchers spanning four continents and supported by Phenomics Australia identified a novel and incredibly rare genetic mutation that represents a new mechanism for human disease.

The challenge:

Seven patients from six unrelated families, scattered across the globe and divided by language, culture, age, and gender, were afflicted with an exceptionally rare genetic mutation. The patients were all very susceptible to infections, and were subsequently experiencing similar symptoms, in fact, they all had infections with one particular pathogen, one that is harmless for most people, but causes severe lung infection for those with serious immunodeficiency.

The solution:

Led by Associate Professor Anselm Enders, from The Australian National University (ANU) Phenomics Australia generated genome edited mouse models though CRISPR/Cas technology to help identify a unique mutation in the IRF4 gene, affecting the immune system's development.

The future:

These insights will allow improved clinical practice through quicker diagnosis and more effective treatment of sufferers of this rare disease.

In Vivo Genome Engineering & Disease Modelling

Producing blood: Researchers zero in on potential new function of lymphatic system

Scientists investigating the causes of lymphoedema have made a major discovery, revealing that lymphatic vessels can produce red and white blood cells. Until now, it was believed that blood cells derived solely from stem cells found in bone marrow.

The Challenge:

Lymphatic vessels are a key component of the cardiovascular system responsible for returning excessive tissue fluid and protein (lymph) back to the bloodstream and form a major part of the immune system that defends the body against harmful bacteria or viruses. Lymphoedema is a condition very difficult to treat characterized by lymphatic system blockages leading to swelling in limbs.

The Solution:

Led by Professor Natasha Harvey from the University of South Australia, and utilising Phenomics Australia genome engineering and histopathology expertise, an international team found that incorrectly programmed genes led to lymphoedema and unexpectedly controlled blood cell production. "If these genes aren't switched on at the correct time and place, lymphatic vessels don't form properly, causing lymph fluid to leak back into the tissues, leading to swelling (lymphoedema). In an unexpected discovery, we identified that the same gene that controls the development of lymphatic vessels also controls the production of blood cells" Prof Harvey says.

The Future:

This groundbreaking discovery suggests that lymphatic vessels could serve as a previously unrecognized source of blood cells during both development and disease. This newfound ability might be significant in infection defense and may have relevance for certain blood cancers.

CRISPR/Cas as diagnostics tools

Phenomics Australia's established genome engineering expertise is used in providing knowledge to look into the commercial and translational potential of CRISPR technology.

The Challenge:

Existing infection tests can take days, or even weeks, to return a result and potentially delay clinical care, pandemic response, and biosecurity threats.

The Solution:

Australian researchers, led by Professor Marco Herold and Dr Andrew Kueh, at the Melbourne Advanced Genome Editing Center (MAGEC) have developed c-FIND, a rapid CRISPR-based diagnostic test that can detect various infections within minutes and it is being transformed into a portable point-of-care device.

The Future:

"c-FIND is fast as well as highly sensitive and specific. This means the technology has the ability to accurately identify if a person has certain infections, as well as rule out the infections a person does not have – it is unique for a test to excel in both these capabilities. Additionally, the system can be easily adapted to test for new and emerging infections," Professor Marco Herold said.

Left: Dr Andrew Kueh at MAGEC laboratory.
Right: Dr Martin Pal at MAGEC laboratory.

In Vitro Genome Engineering & Disease Modelling

Using miniature brain models to prevent developmental impairments

Phenomics Australia scientific leader Professor Wolvetang is renowned for his work at the Australian Institute for Bioengineering and Nanotechnology (AIBN) in organoids, tiny, three-dimensional avatars that are grown to resemble real human organs.

The Challenge:

Birth asphyxia (HIE) affects four in 1000 newborns, leading to severe disabilities and a high mortality rate of up to 60% in affected infants, with limited treatment options for preventing long-term impairments.

The Solution:

Professor Wolvetang and his team received a \$1.36 million grant from the NHMRC to use brain organoids to study HIE and develop better treatments for affected infants, aiming to improve early diagnostics and therapeutics.

The Future:

By studying brain organoids and the various HIE stages, this research may revolutionize the understanding of HIE's acute and long-term effects. "Through this work, we are fostering the development and testing of new individual therapies which when applied at scale, could have a huge global health and economic impacts."

ISSCR Launches Standards for Human Stem Cell Use in Research

The International Society for Stem Cell Research (ISSCR) has released the ISSCR Standards for Human Stem Cell Use in Research, an international collaboration aimed at enhancing rigour in preclinical research and ultimately strengthening the pipeline of therapies for patients.

The Challenge:

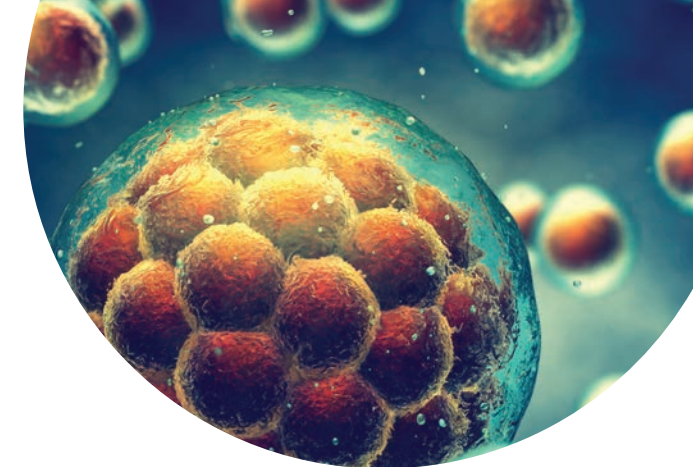
Ensuring the rigour and reproducibility of stem cell research is essential for advancing preclinical research and developing therapies for patients.

The Solution:

Phenomics Australia scientific director and Stem Cell research expert Professor Christine Wells was part of this international panel of experts that launched the ISSCR Standards for Human Stem Cell Use in Research. These standards provide guidelines for publishing stem cell research, improving reproducibility, and facilitating data submission to leading stem cell journals.

The Future:

These standards establish quality criteria and core principles for working with human stem cells, enhancing research quality and data reporting in basic research laboratories, ultimately advancing the field of stem cell research.



Functional Genomics & High-Throughput Screening

Probing the protein factories of the cell to treat rare diseases.

The Challenge:

Proteins, crucial for cell survival and growth, depend on ribosomes for synthesis. Disruptions in ribosome biogenesis, caused by deficiencies in ribosomal proteins and ribosomal RNA, lead to ribosomopathies like Diamond-Blackfan Anemia (DBA), which can result in bone marrow failure and anemia requiring lifelong clinical management.

The Solution:

Using state-of-the-art robotic and liquid handling facilities in two Phenomics Australia nodes in the ACTD and the Victorian Centre for Functional Genomics (VCFG) at the Peter MacCallum Cancer Centre, Dr. Amee George's team used functional genomics to identify genes that control the cellular p53 protein, a key factor in the stress response leading to ribosomopathies.

The Future:

Understanding the molecular mechanisms behind ribosomopathies and identifying therapeutic targets could lead to groundbreaking treatments for DBA and similar conditions. This research may bring life-changing discoveries and improved treatments especially to individuals with limited therapeutic options.

New treatment for acute myeloid leukaemia could prove beneficial for even more people.

New research published in Science Advances conducted by researchers at Peter MacCallum Cancer Centre and supported by Phenomics Australia identified a potential new treatment for two challenging blood cancers that could help more patients than originally thought.

The Challenge:

Finding effective treatments for acute myeloid leukaemia (AML) and myelodysplastic syndrome (MDS), especially beyond patients with the TP53 mutation, has been challenging.

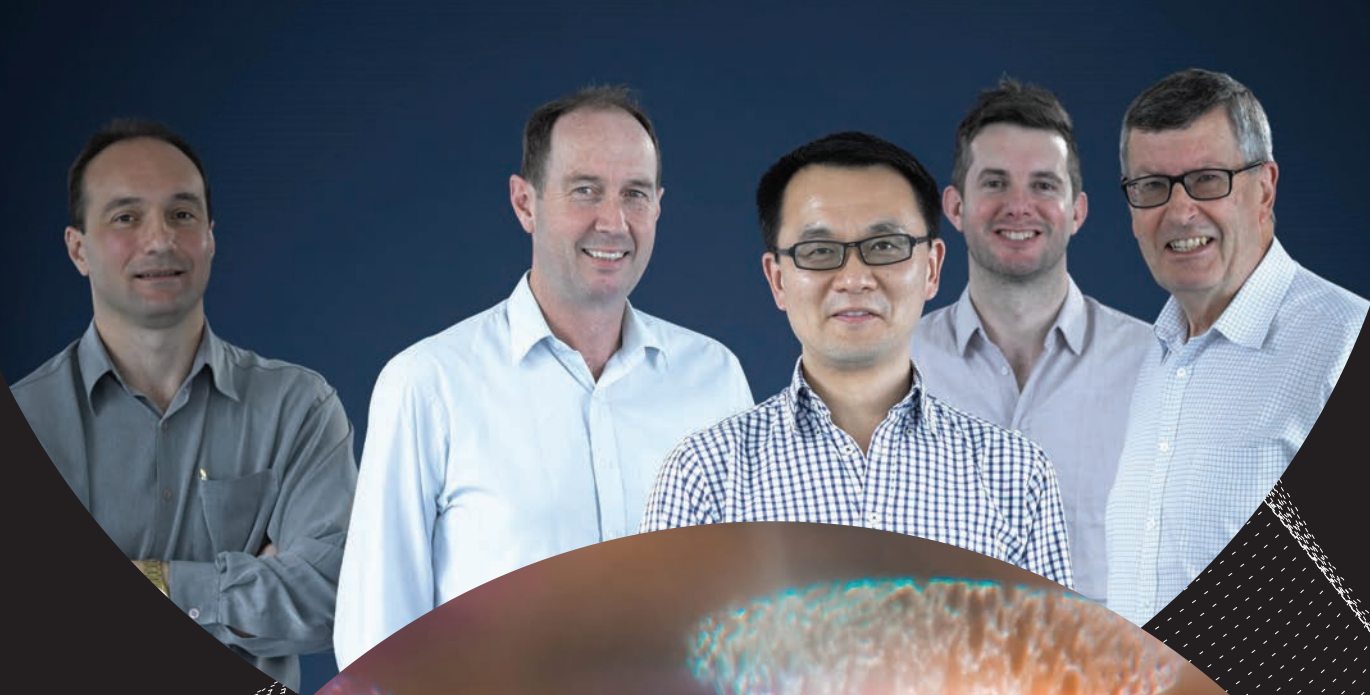
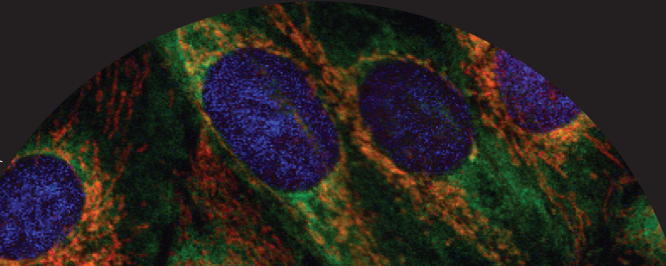
The Solution:

Researchers at Peter MacCallum Cancer Centre used a genome-wide CRISPR screen with support from the Phenomics Australia node at the Victorian Centre for Functional Genomics (VCFG) to discover that the treatment eprenetapopt may have a broader range of effectiveness, potentially benefiting more AML and MDS patients, especially those accumulating iron.

The Future:

Clinical trials for this treatment are underway, offering hope for a wider group of patients in need of new treatment options. Researchers also aim to explore its potential application in other cancers beyond AML and MDS. This research opens up possibilities for more effective treatments in the future.

Top centre: L-R Associate Professor Tony Velkov, Professor Philip Thompson, Professor Jian Li, Dr Kade Roberts, Professor Roger Nation. Image credit Monash University



Pathology

New antibiotic candidate to tackle deadly bacterial 'superbugs'

The Challenge:

Sepsis, a deadly bacterial infection, affects more than 55,000 Australians annually and is a global health crisis. Infections from drug-resistant Gram-negative 'superbugs' pose a serious threat, with dire consequences if not effectively treated. Without new antibiotics, antibiotic resistance could lead to 10 million deaths each year by 2050.

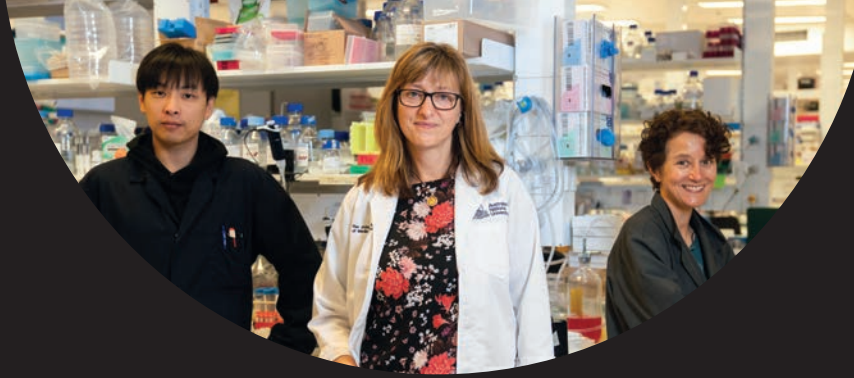
The Solution:

Researchers from Monash University, led by Professor Jian Li and with the support of Phenomics Australia, have developed QPX9003, a novel antibiotic targeting drug-resistant Gram-negative bacteria. This antibiotic is in Phase 1 clinical trials, marking significant progress in addressing this urgent healthcare challenge.

The Future:

The long-term research collaboration of Professor Li and his team utilising the unique services offered by the Phenomics Australia Histopathology and Digital Slide service for their antibiotic discovery project will continue to explore innovative antibiotic candidates to combat drug-resistant bacteria. These efforts are crucial in averting a potential global health catastrophe and economic burden caused by antibiotic resistance.

Top right: Senior Lecturer Dr Vicki Athanasopoulos (C) with Researcher Dr Hao (Howard) Wang (L) and Senior Technical Officer Jean Cappello (R). Image: Tracey Nearmy/ANU



Paving the way for new and tailored lupus treatments

Phenomics Australia infrastructure, including the Genome Engineering service at the Australian National University, the Australian Phenomics Facility and the Histopathology and Slide Scanning service at the University of Melbourne played a long-term collaborative role in this study.

The Challenge:

Lupus, a chronic autoimmune disease, lacks a cure and current treatments have side effects. Researchers aimed to unravel the genetic basis of lupus, paving the way for more effective treatments without adverse effects.

The Solution:

An international team conducted whole genome sequencing on a child named Gabriela, who had severe lupus with an early onset, identifying a single genetic cause—a mutation in the TLR7 gene. Using CRISPR gene-editing, they confirmed the mutation's role in lupus by introducing it into mice, which developed similar symptoms.

The Future:

This groundbreaking discovery may lead to new drugs targeting the TLR7 gene, potentially revolutionizing lupus treatments. The researchers are collaborating with pharmaceutical companies to explore therapeutic development or repurposing. Additionally, the mouse model will aid in testing therapies for lupus patients, offering hope for better treatments and reduced side effects.



Beyond Health

World first technology to suppress invasive mice

Researchers at the University of Adelaide, including Phenomics Australia Scientific Director and CRISPR expert Prof. Paul Thomas, have released their first findings on the potential effectiveness of revolutionary gene drive technology to control invasive mice.

The Challenge:
Invasive mice pose a significant environmental and economic challenge, and finding an effective, humane, and environmentally friendly way to control their populations is vital.

The Solution:
Researchers at the University of Adelaide have developed t-CRISPR technology, using cutting-edge DNA editing technology to make alterations to a female fertility gene. Once the population is saturated with the genetic modification, all the females that are generated will be infertile. "We are also developing new versions of t-CRISPR technology that are designed to target specific pest populations to prevent unwanted spread of the gene drive." Prof Thomas said.

The Future:
The research offers a promising solution for managing invasive species, with the potential to benefit the environment, communities, and agriculture. The researchers are working on strategies to address gene drive resistance, collaborating with organizations like CSIRO and considering societal views and attitudes towards this technology. This innovative approach could pave the way for similar solutions for other invasive pest animals.

Preventing Mastitis in Dairy Cows

Phenomics Australia histopathology services have been applied to develop techniques to prevent mastitis in non-human mammalian subjects, such as dairy cows.

The Challenge:
Mastitis (inflammation of the mammary gland or udder) is the single most costly disease in the dairy industry costing the US dairy industry about US\$2 billion annually or 11% of the total US milk production.

The Solution:
A patented method of reducing or shutting down lactation in a non-human mammalian subject by administering to the subject by intramammary infusion an agent that activates the OAS2 signalling pathway or which induces expression of OAS2.

The Future:
The methods and reagents described in the patent can potentially mitigate a significant and costly risk to agricultural productivity, not only in the dairy industry but possibly other animal species.

Nematode Control of Livestock

Phenomics Australia node at the Victorian Centre for Functional Genomics (VCFG) has performed compound screenings that enabled the repurposing of known drugs to control parasitic nematodes in livestock.

The challenge:
The control of gastrointestinal nematodes in livestock has relied largely on the use of a limited number of anti-parasitic drugs. However, drug resistance is now very widespread and no vaccines are available for the vast majority of these parasites

The solution:
The development of new drugs is crucial to ensure effective and sustained nematode control in the future.

The future:
The repurposing of known drugs or bioactive chemicals with known safety profiles and modes of action has the potential to proceed to clinical trials more rapidly than newly discovered chemotypes.

Supporting bushfire research, recovery and resilience

The Challenge:
Understanding the health impacts of bushfires and air pollution on human health is crucial in the face of increasing climate-related challenges in Australia.

The Solution:
Phenomics Australia's research support encompasses issues like air pollution exposure during pregnancy, respiratory diseases, and ocular diseases providing expertise and customized model systems to support the effects of bushfires and air pollution on human health.

The Future:
Investments in world-class climate and environmental science are essential to address the challenges posed by climate change and environmental shifts. Environmental monitoring, pollutant source management, and response strategies for bushfires and dust storms will help safeguard communities, industries, and ecosystems, ensuring better adaptation to climate threats.

Our Team

Phenomics Australia Board

The Phenomics Australia Board provides oversight and strategic guidance for all Phenomics Australia activities and investments. The Board members provide extensive experience in scientific strategy, biomedical research, policy, governance, industry engagement and commercialisation.

- Dr Liz Jazwinska** – Chair
Dr Carol Wicking
Dr Susan Pond
- Prof. Ute Roessner**
Dr Michael Poidinger
Prof. Glenn Withers

Phenomics Australia Scientific Leaders

The Phenomics Australia Scientific Leaders are responsible for the delivery of Phenomics Australia's various services, while regularly providing highly informed scientific, technical, and strategic advice to the Board.

- Prof. Kaylene Simpson**
Head Of Victorian Centre For Functional Genomics (VCFG) – Peter MacCallum Cancer Centre
- Dr Andrew Kueh**
Head Of The Melbourne Advanced Genome Editing Centre (MAGEC) – Olivia Newton John Cancer Research Institute
- Prof. Paul Thomas**
Head of the South Australian Genome Editing (SAGE) Facility – South Australian Health and Medical Research Institute (instead of SAHMRI)
- Dr Alexander Combes**
Head Of The Monash Genome Modification Platform (MGMP) – Monash University
- Prof. Janet Keast**
Head Of The Histopathology And Digital Slide Service – University Of Melbourne



Dr Louise Winteringham
Head Of The Translational Cancer Research Program – The Harry Perkins Institute of Medical Research

Prof. Graham Mann
Director Of The John Curtin School Of Medical Research – Australian National University

A/Prof. Amee George
Manager of the ANU Centre for Therapeutic Discovery (ACTD) – Australian National University

Dr Thierry Jarde
Director of the Monash Organoid Program – Monash University

Dr Sara Howden
Head of the Induced Pluripotent Stem Cell (iPSC) Derivation & CRISPR Gene Editing Facility – Murdoch Children's Research Institute

Prof. Christine Wells
Chair of Stem Cell Systems – University of Melbourne (UMelb)

Prof. Alice Pébay
Head of the Stem Cell Disease Modelling Laboratory – University of Melbourne

Prof. Ernst Wolvetang
Head of the Stem Cell Engineering Laboratory, Australian Institute for Bioengineering and Nanotechnology – University of Queensland

Prof. Sally Dunwoodie
Stem Cell Production and Cell Function Screening Service – Victor Chang Cardiac Research Institute

A/Prof. Sefi Rosenbluh
Head of the Monash Functional Genomics Platform (MFGP) – Monash University

Prof. Simon Barry
Head of the Functional Genomics South Australia (FGSA) – University of Adelaide

Prof. Kieran Harvey
Head of the Australian Transgenic Drosophila Facility – Peter MacCallum Cancer Centre

Phenomics Australia Executive Team

The Executive Team is responsible for the day-to-day operation of Phenomics Australia.

- Prof. Michael Dobbie**
Chief Executive Officer
- A/Prof. Jim Hennessy**
Chief Operating Officer
- Dr Marina Trigueros**
Communications and Outreach Coordinator
- Dr. John Parisot**
Strategic Partnerships Advisor
- Ms Sarah Dreese**
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- Dr Twishi Gulati**
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