

Melbourne Genomics
Health Alliance

Revolution becomes routine

Annual Report 2023



Alliance members



Supported by



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Understanding the genetic cause of Edward's kidney disease enabled his doctors to know how the condition will progress and what preventive steps to take. Photo: Melbourne Genomics/Rodney Dekker

The story so far

Genomic medicine uses the big data in our cells to diagnose illness and find the right way to prevent or treat it.

In 2013, leading hospitals, research and academic institutions joined forces to create the **Melbourne Genomics Health Alliance**. Their shared goal was to make genomics a routine part of healthcare, so patients could benefit.

The Alliance made Victoria the first place in the world to provide real-time evidence of how genomics could improve patient care. More than 4,000 health professionals gained critical skills in genomic medicine. A world-first clinical genomic data platform has enabled six medical laboratories to provide more than 22,000 genomic tests for Victorian patients.

The Victorian Government has supported and championed this work for more than a decade.

Now the Alliance is in the final years of its final program. When it ends, thousands of Victorian health professionals can continue to take genomics forward, equipped with 12 years' worth of evidence, education and resources.

The year in numbers

Patient care



13 health services involved in projects that support the use of genomics in patient care.



29 clinicians funded to tackle barriers to the wider use of genomics in Victoria.

Education



842 healthcare and science professionals and students accessed Melbourne Genomics education.



97% of doctors were more confident in at least one genomic skill after clinical education.



100% of Victorian health regions represented in the health professionals who attended clinical genomics education.

Community input



34 community members informed and shaped genomics projects.

Technology



The Genomical software platform has supported **more than 22,000 accredited genomic tests** across six medical laboratories.



This year saw a **40% increase** in genomic tests conducted through Genomical.

Value

These figures represent the cumulative value of the current program since 2020.



\$66.1M leveraged since 2020.

End-of-program target: \$38.9M (exceeded).



6.5M generated through MBS item usage.

End-of-program target: \$5.25M (exceeded).



31.55 FTE in new jobs outside the program.

End-of-program target: 24 FTE (exceeded).

Report from the Chair of the Board

Catherine Walter AM

In November 2023, 84 guests joined us at the State Library to celebrate a decade of genomic medicine in Victoria.

Some were leaders and clinicians from renowned hospitals. Others were pioneering scientists in various fields. There were educators and academics, policy advisors and software developers, as well as healthcare consumers who had become passionate advocates for genomics.

That night, we reflected on what had enabled some of the big changes of the past decade: from real-time evidence of the benefits of genomics, to new Medicare items for genomic testing, new genomics facilities, thousands of doctors and scientists trained, and so on.

It was wonderful to hear Professor Doug Hilton recall the discussions that led to the Melbourne Genomics Health Alliance being formed – and why collective action from research, healthcare and academia was needed. Doug has been involved in the Alliance from the very beginning: first as head of WEHI and now as CEO of CSIRO.

Professor Shelley Dolan's observations were equally insightful. As CEO of The Royal Melbourne Hospital, she discussed the challenges of working within a complex health system and commended the Alliance for working with hospitals to design fit-for-purpose solutions.



Photo: Melbourne Genomics/Thomas Elia

Now the Alliance is in its final program. Once it has concluded, it will be up to each member organisation – alongside many others across Victoria – to take genomics forward.

The Board's focus is firmly on the sustainability of our shared endeavour. We decided to spin out a commercial venture to ensure the Genomical® data platform remains available to Alliance members and other laboratories that need it for genomic testing. The new company is wholly owned by the Alliance members.

We also recognise it will take collective effort from the Victorian Government and healthcare providers to make genomics part of usual patient care. This will be the core of our advocacy in the year ahead.

On behalf of the Board, I wish to thank The Hon Mary-Anne Thomas, The Hon Ben Carroll, and the Departments of Health and Medical Research for their ongoing support.

Report from the Executive Director

Professor Clara Gaff

Ten years ago, Melbourne Genomics was formed to bring genomic medicine into Victoria. Two years from now, it will end as always planned.

We knew back then that genomics would revolutionise patient care – but as author and comedian Bassam Youssef points out: "A revolution is not an event. It is a process." Thus, the Alliance members together embarked on a staged process to bring genomic testing into Victoria's complex, adaptive and decentralised healthcare system.

Our projects have always explored real-world barriers and solutions. This year, we invited hospitals across the state to join us in co-creating a clinical governance framework for genomics. This triggered valuable conversations between the clinicians who are championing this emerging field of medicine, and the hospital leaders responsible for ensuring it can be provided safely and effectively.

We gave revolutionary ideas space to grow. This year, brilliant clinicians explored ways to support their medical and nursing colleagues to use genomics, bring genomics into their specialities, and even road-test new uses for genomics. In doing so, they are informing models of care that are ideally suited to Victoria.



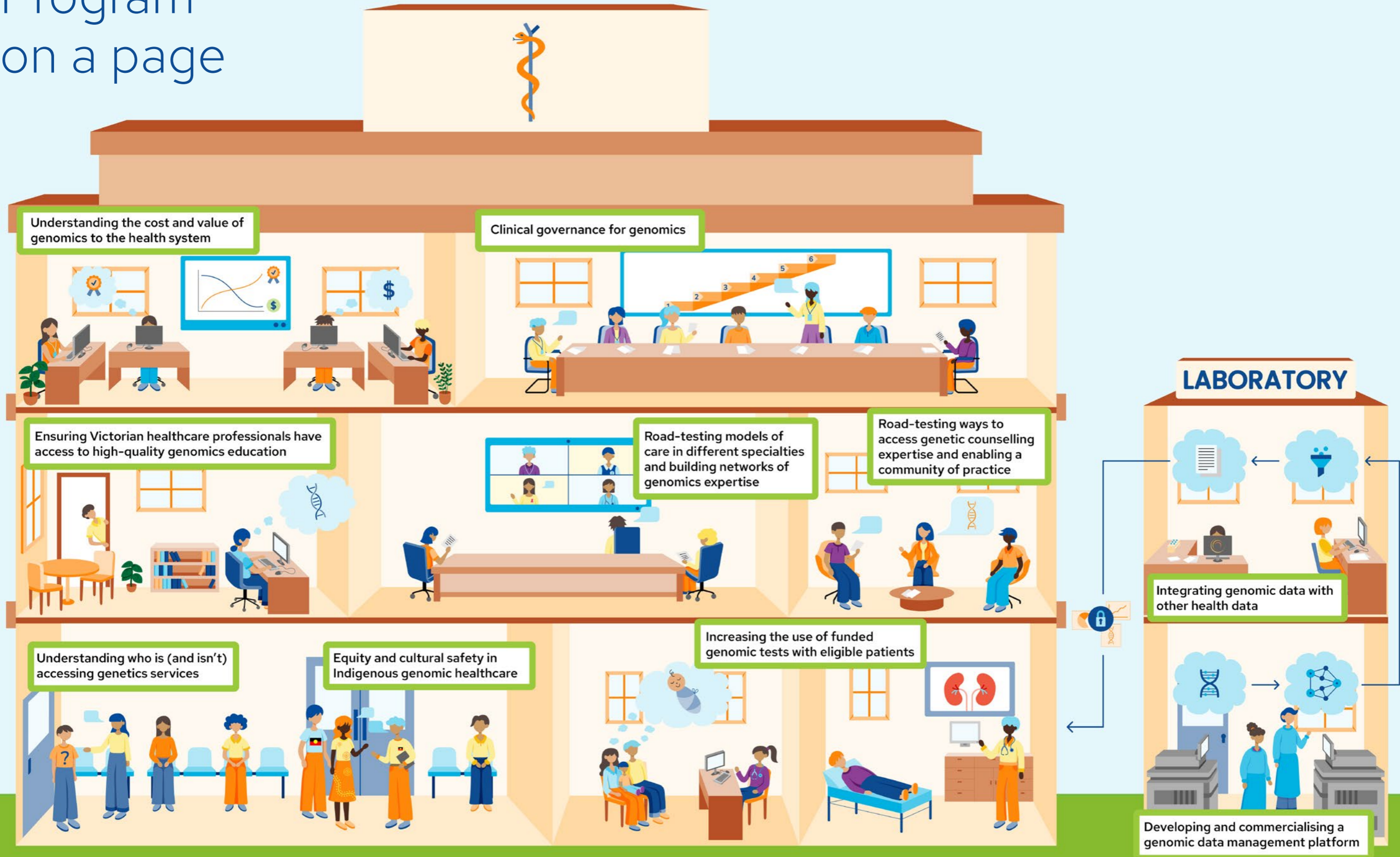
Photo: Melbourne Genomics/Thomas Elia

Our Victorian alliance is just one arm of a broader movement across the country. We stand alongside other genomics consortia, patient support groups and industry bodies, all advocating for the equitable use of genomics in healthcare.

The formation of the Australian Alliance for Indigenous Genomics (ALIGN) was a significant step towards equity. We were honoured to provide administrative support to the Victorian node of ALIGN, and to partner with VACCHO to explore how genomic medicine can equitably benefit Aboriginal and Torres Strait Islander peoples in Victoria.

As the Melbourne Genomics program works towards its planned end, I am heartened by the number of families and communities, doctors and scientists, researchers and policymakers who are now ready to take genomics forward.

Program on a page



Progress report



Diagnosis Day followed six families with rare genetic conditions. Read more on page 16.
Photo: Melbourne Genomics/Rodney Dekker

Co-designing a revolution

If it doesn't exist, build it together

Clinical governance frameworks help hospitals make good decisions about safe and effective care. But none currently address a field as encompassing and evolving as genomics – so a new framework was needed.

Victorian healthcare leaders across and beyond the Alliance collaborated on a world-first tool to help hospitals assess their capability to provide genomic medicine safely and effectively.

Grounded in familiar clinical governance domains, the tool addresses considerations specific to genomics. Hospitals can check their progress, determine how best to ensure their patients can access genomic medicine, and plan their next steps.



70 healthcare professionals contributed to building and testing the tool in 2023.



5 hospitals are involved in usability testing.

“The tool promoted thinking about different aspects of service delivery, and how request, store, implement and track genomic requests and results within our health service ... It is useful to prompt thinking about areas that we, as a service, need to improve upon – and then start the conversation about how those improvements can be achieved.”

– Dr Patricia Banks, Medical Oncologist, Barwon Health



Many paths to the same destination

Several Melbourne Genomics projects road-test ways to bring genomics into specialty areas. This will inform future models of care.

Genetic counsellors are key to many of these projects. Some are embedded within speciality clinics, others provide support by phone, during scheduled meetings, or onsite (see diagram opposite). Research is underway to understand how genetic counsellors can best support medical specialists to use genomics in patient care.

The **Cancer Statewide project** is trialling three ways for patients to access genomic testing and get their results. Six regional and metropolitan hospitals are participating in the trial.

Models for providing genomics in cancer care



Genomic testing and results provided by on-site oncologists.



Referral of patients to an expert site to discuss genomic testing and result implications, conducted via telehealth.



Local oncologists are trained as genomics 'superusers' to support test delivery at their own hospital.

The **Nephrology Clinical Change project** has developed a traffic-light system to help nephrologists identify which of their patients may benefit from genomic testing. Complex cases are discussed at a monthly meeting of genetics and nephrology experts.

The **Dementia Clinical Change project** is exploring how to provide timely access to genomics for people with early-onset dementia and cognitive disorders. Genetic counsellors now work within neuropsychiatry and neurology clinics at four hospitals, supporting patients and their blood relatives to get tested for genetic causes of dementia.

What genetic counsellors do in our projects



Manage genetic care for patients after return of test results



Advise clinicians about conducting consent discussion



Provide clinician education



Manage a support line and email



Provide genomic testing and pre- and post- test counselling to patients



Advise clinicians through multidisciplinary team meetings



Co-conduct research with project leads



Facilitate genomic testing



Advise clinicians about patient triaging



There are many ways for patients to get high-quality genomic healthcare, close to home. We are working with hospitals to road-test different models of care. Photo: Melbourne Genomics

Innovating on the frontline

The **Cancer Statewide project** is testing out how a new technology can be implemented in patient care.

Cell-free tumour DNA (ctDNA) screening analyses small particles of cancer DNA in a patient's blood sample, without requiring a biopsy of the tumour. Six Victorian hospitals are participating in the project, led by the Peter MacCallum Centre.



ctDNA testing

Enables cancer DNA particles to be analysed from a blood sample, a non-surgical option that is easier to organise and pain-free for patients.

Meanwhile, the Transplant Clinical Change project is investigating how genomic testing can inform the care of people who will receive kidney and liver transplants. Experts in transplant

surgery, pathology, genetics, bioinformatics, and pharmacy created a 'genomic x-ray' – a snapshot of genomic insights that can inform diagnosis and guide treatment options.

The team has already uncovered previously unknown diagnoses and identified individuals with rare responses to medications.



'Genomic x-ray' report

The human genome contains a vast amount of information to guide patient care. This report is for people who need organ transplants, and will synthesise genomic information about risk factors, co-morbidities and potential adverse reactions to anaesthesia and other medications, to provide each patient with personalised care.

Changing hearts, minds and practice

Knowing when and how to use genomics

Some genomic tests are still under-utilised, even when Medicare funding is available.

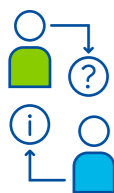
The **Paediatrics Statewide** and **Nephrology Clinical Change projects** are exploring how best to support doctors to use genomic testing for patients who need it.



Step-by-step guides

The websites paediatricgenomics.org.au and kidneygenomics.org.au walk clinicians through the test ordering process, providing decision support tools and links to eligible pathology providers.

Resources on these sites have been used **more than 950 times** in 2023 to support decisions about genomic testing for patients.



'Ask an expert' service

Dedicated phone lines connect Victorian paediatricians or nephrologists with genetic counsellors who have expertise in their speciality.



Local champions

Both projects are led by doctors with expertise and networks, who act as peer leaders in their discipline. The Nephrology project centres on 'genomics champions' who can support their colleagues at each participating hospital.

Evaluating these projects can show what interventions are most effective in changing practice.

Giving clinicians a confidence boost: anytime, anywhere

As genomics becomes more and more mainstream, demand for quality genomics education will continue to grow.

Evaluation has shown **97% of doctors** who completed Melbourne Genomics courses felt more confident about using aspects of genomics in their practice.

In late 2023 we tested out wholly online courses, enabling doctors to learn genomics anytime, anywhere there's internet. **186 professionals** have accessed the courses since they launched in October 2023. The limited time offer was extended into 2024 due to popular demand.

We are now exploring options to make up-to-date genomics education available to clinicians beyond the life of our program.



842 health professionals built their genomics expertise as a result of our education.



97% of doctors felt more confident in aspects of genomic medicine after clinical education.



186 professionals have accessed our online courses since they launched.

Time and space to solve problems

When doctors and scientists get space and time to think about the systems they operate in, they can drive an astonishing amount of change.

Our Implementation Fellowships enabled 17 clinicians and scientists to progress the use of genomics in healthcare. Their work ranged from supporting cardiologists to use genomic testing, to preparing laboratories to scale up and meet demand, to bringing genomic testing into maternal and foetal medicine. (See **page 26** for a full list of Implementation Fellowships.)

'DNA detective' is a career of choice

Demand for **variant interpretation** education is at an all-time high – from university students to well-established medical scientists.

Our online modules – which delve into the detective work of analysing genetic variants for clinical care – were developed for introductory and advanced levels.

The Australasian Society of Diagnostic Genomics has licensed access to these modules and will continue to make them available to medical scientists who want to build a career in genomics.

Meanwhile, Melbourne Genomics teamed up with Melbourne Bioinformatics, InGeNA and The University of Melbourne to host three careers events, attended by a total of 187 students and professionals from medical and data science. There was strong interest in diagnostic genomics as a career option.



Patients drive the revolution



Patient perspectives change everything

Our Community Advisory Group recommended the formation of project-specific consumer panels, to ensure people with lived experience could inform genomic medicine services for others like them.

Three new consumer panels were established during the year. They have already:

- advised on how genomic test results are communicated to people with cancer
- developed patient-friendly information on genomic testing in organ transplant care
- discussed appropriate ways to talk to families about genetic causes of dementia.



32 community stakeholders contributed to the program in 2023.

Finding patterns in the numbers

Genomics should be for everyone, but will some people miss out while others get the best of care?

A study led by The University of Melbourne aims to find gaps in service, so they can be addressed before genomics goes fully mainstream.

The study uses data from genetics services, obtained through the Victorian Agency for Health Information (VAHI). Delving into this data can show whether Aboriginal and Torres Strait Islander patients in Victoria are accessing genetics services at expected rates – and if not, whether the challenges lie in referral, initial appointments, or specific types of genetic healthcare.

Similar analysis will be performed to see whether access patterns are similar or different. Both analyses can help service providers identify barriers or areas to strengthen.

First Nations leaders shape the future of genomics

Melbourne Genomics was proud to support the **Australian Alliance for Indigenous Genomics (ALIGN)** in establishing a Victorian presence and Indigenous Governance Committee.

Led by First Nations researchers, health workers and community leaders, ALIGN works to ensure Aboriginal and Torres Strait Islander peoples explicitly benefit from genomic medicine and have sovereignty over their genomic data.

Collaborating with ALIGN opened the door to a new Melbourne Genomics funded project led by the **Victorian Aboriginal Community Controlled Organisation (VACCHO)**. This project explores barriers and access to genomic medicine and genetic service referrals, from the perspectives of Aboriginal health services and communities.

The project began in November 2023, with strong ethical and Indigenous governance frameworks now in place.

How patients feel about storytelling

Diagnosis Day was a storytelling project about rare genetic conditions, produced by Melbourne Genomics and the Genetic Support Network of Victoria.

An evaluation of the project found the videos had high engagement, and that people who shared their stories felt respected and supported. The evaluation report is available at diagnosisday.org.au and contains insights on working collaboratively, recruiting participants, participant experience and distribution of stories.

Genomic testing discovered the ultra-rare reasons for Charlie and Rosie's mystery illnesses.

What happened to us was a bit like lightning striking multiple times.

Charlie, our eldest, was born with a congenital heart condition. He had ongoing breathing difficulties, which his doctors thought was the result of the heart surgery he had just after birth. A simple childhood illness could see him back in hospital on oxygen for weeks at a time.

Meanwhile, Rosie was a very healthy baby until she became unwell at 10 months old. Over two weeks she lost weight, lacked energy and was pale – symptoms which took us to the emergency department just in time to save her life – she was in acute heart failure. Her doctors diagnosed dilated cardiomyopathy, but the gene panel tests they did at the time couldn't explain why. A whole exome test found Rosie has a rare mitochondrial mutation that Sinead (her mum) shares, which can affect her heart and other organs.

We moved from London to Melbourne last year, and our kids have had great care at The Royal Children's Hospital. Rosie has continued to improve with only a few protective medications. Charlie on the other hand, was struggling to put weight on and his growth rate was dropping off. He then developed persistent chilblains over the winter and random and severe joint pains. His constellation of symptoms was worrying and unexplained, until we were referred to the Monash Health genetics team.

They performed a whole exome test which identified an incredibly rare autoimmune condition caused by a mutation on the STING1 gene. **The difference that test made is unfathomable.** Not only was Charlie spared biopsies and other invasive tests, but his doctors

know that the standard immunosuppressive treatments don't work for his condition – so he receives a targeted medication that has helped significantly improve his quality of life and life-expectancy.

Charlie's an amazing, resilient kid – he's exploded with confidence recently and wants to play football even when he's in pain. Rosie is feisty and loud and imaginative. Both of them will need lifelong medication, ongoing monitoring and care, but the doctors know what to look for. Life is good now – we don't quite know what the future holds but we're trying to live in the present.

Sinead and Jon

Photos: Melbourne Genomics/Rodney Dekker



The right technology to scale genomics

Genomical® set to reach the stars

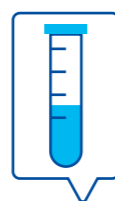
From 2016–2020, Alliance members built **GenoVic**: a software platform that could manage the end-to-end workflow of a genomic test and securely store genomic data in the cloud.

From 2020–2022, we put the platform to work in several accredited laboratories, demonstrating how it could save hours of manual work per test and enable pathology providers to meet the growing demand for genomic testing.

In 2023, it was time to make the platform self-sustaining. We engaged users, investors and genomics experts in a consultative process to identifying key selling points and a new name.

GenoVic became **Genomical®**: a name that conveys the size and complexity of the data it can manage.

Note: A new commercial venture called Transcendomics was formed in 2024 to ensure the Genomical platform remains available to laboratories across and Victoria, even after the Alliance's programs conclude.



Genomical has supported **more than 22,000 accredited genomic tests** across six medical laboratories.

The number of tests done through Genomical increased by **40%** this year.

Some of the Melbourne Genomics team who helped build the Genomical platform. Photo: Melbourne Genomics.



Genomical helps laboratories to scale up genomic testing. Photo: iStock

“With Genomical, it takes half the time to do what we need to. Everything happens automatically in the background, with data going straight from the sequencer to our analysis tools. We don’t have to do this manually anymore. Genomical is a one-stop shop for all of the data complexities we were dealing with, from data management to storage and access.”

- Dr Ravikiran Vedururu, Genetics and Molecular Pathology Laboratory, Monash Health



Genomical.

Your launchpad for genomic medicine.

The value of a vendor-agnostic platform

Genomical’s advantage became rapidly apparent when a widely used tertiary analysis tool, Alissa Interpret, was withdrawn from the market. Many medical laboratories across the country relied on this tool for variant interpretation – the most intricate step in a genomic test.

Genomical is a platform for multiple tools, all used for different types of tests, or for different stages in a genomic test. Its support team was rapidly able to identify alternative tertiary analysis tools. In 2024, the team will work with each lab to implement their preferred tool. This demonstrates the platform’s value in enabling genomic experts to maximise time on patient care rather than lengthy and costly procurement and IT integration activities.

Human blueprints need protection

Genomic data is highly personal and sensitive. It needs strong protections, especially in light of recent hacking attempts on health and government services.

This year, Genomical’s compliance was confirmed with the Victorian Department of Health’s cybersecurity framework, the Victorian Privacy Act and Australian Privacy Principles. Ongoing privacy assessments, security reviews and training ensure that Genomical and its users can stay ahead of the latest cybersecurity threats.

Turning good ideas into sound solutions

Genivate 23 is a fund that helps Alliance members road-test digital solutions to genomic testing challenges.

Scientists were invited to submit proposals to test new technologies and data management approaches within their own laboratories.

Looking back, looking ahead

Recognising a decade of progress

October 2023 marked 10 years since the Melbourne Genomics Health Alliance was formed.

We celebrated with a **report** that chronicled 10 years of genomic advancements in Victoria – from the first real-time evidence for the use of genomics in patient care, to new Medicare items for genomic testing, to a world-first genomic data management platform and beyond.

The report was shared in a media release by Ministers Mary-Anne Thomas and Ben Carroll, and featured in a story in the Herald-Sun.

On 3 November 2023, we threw a party at the State Library of Victoria to celebrate progress, collect stories of impact, and look to the future.

“It typically takes an average of 17 years for research evidence to reach clinical practice, meaning the achievements of the Melbourne Genomics Health Alliance in just 10 years are nothing short of remarkable.”

- The Hon Mary-Anne Thomas, Minister for Health

“Genomics has become essential to a growing number of medical specialties. What we need is the collective will and effort to make it happen. The future is ours. The future is Victorian.”

- Prof Shelley Dolan, CEO of The Royal Melbourne Hospital



Enabling genomics research to flourish

Over more than a decade, the work of Melbourne Genomics has helped cultivate an environment where genomics research can flourish. This research now brings federal funding into Victoria, through the Australian Government's Medical Research Future Fund.

In 2023, more than \$21M was granted to projects involving Melbourne Genomics member organisations.

The University of Melbourne received funding for two projects on cancer and polygenic risk scores, one of which is led by Professor Paul James. The project is trialling the integration of polygenic scores for common cancers alongside current genetic testing in standard clinical care.

The Murdoch Children's Research Institute received three genomics grants, investigating the implementation of pharmacogenomics for children and the rapid diagnosis and tailored management of infantile epilepsies. A third project led by Professor Paul Lockhart is exploring the use of new technologies for diagnosis of ataxia and repeat expansion disorders. It builds on the work of the Melbourne Genomics Complex Neurological and Neurodegenerative Diseases Clinical Project.

Dr Vanessa Bryant and Dr Charlotte Slade – who had previously led a Melbourne Genomics immunology project – were confirmed as chief investigators on a new project using a multi-omics approach to expedite diagnosis and management of immune diseases.

Associate Professor Belinda McClaren will be working on a project using implementation science to design a robust national approach to embedding genomics in primary care. Additionally, Professor Sue White and Professor Tiong Tan are part of a team establishing a national long-read genome sequencing program to improve rare disease diagnosis.

What's worked so far? And what needs new thinking?

A big part of creating change is evaluating your progress. We reviewed the Alliance's current program at its halfway mark, gathering insights to inform our final years.

The **formative evaluation** shows that co-design and collaboration are key to success; that sustainable change comes from within the system; and that we need to provide more opportunities for healthcare leaders and genomics experts to connect. The Alliance's purposeful, collective and staged approach to bringing genomics into healthcare was seen as unique by its stakeholders.

But the work is far from done. Knowledge mobilisation is the critical next step: turning evidence into practice, identifying people with the expertise, power and will to take genomics forward, and ensuring that decision-makers see genomics as essential for safe, effective healthcare.



The people taking genomics forward

Board of Directors

The Alliance's Board comprises leaders from each of the 10 member organisations, and an Independent Chair.

The Board has two committees: Finance, Audit and Risk; and Business Development and Commercialisation. Four Board meetings were held in 2023, with committee meetings held beforehand.

The shared investment and mutual commitment from the members makes the Board a strong collective voice for genomics.

Board of Directors

Catherine Walter AM

Independent Chair
Appointed Jul 2016

Prof Christine Kilpatrick

Chief Executive
The Royal Melbourne Hospital
Appointed Oct 2015
Retired from Board Jun 2023

Bernadette McDonald

Chief Executive Officer
The Royal Children's Hospital
Appointed Sep 2021

Prof Mike McGuckin

Associate Dean Research, Faculty of Medicine
Dentistry and Health Sciences
The University of Melbourne
Appointed Mar 2018

Prof Doug Hilton

Institute Director
WEHI
Appointed Oct 2015
Retired from Board Aug 2023

Carolyn MacDonald

Chief Operating Officer
WEHI
Appointed Sept 2023

Prof Kathryn North AC

Director
Murdoch Children's Research Institute
Appointed Oct 2014

Lynne Cobiac

Director (Acting) Health and Biosecurity
CSIRO
Appointed Sep 2022
Retired from Board Dec 2023

Joe Baini

Chief Executive Officer
Australian Genome Research Facility
Appointed Dec 2021

Prof Shelley Dolan

Chief Executive Officer
Peter MacCallum Cancer Centre
Appointed Dec 2019
Retired from Board Jun 2023

Chief Executive Officer
The Royal Melbourne Hospital
Reappointed to Board Sep 2023

Prof Jason Payne

Chief Executive Officer
Peter MacCallum Cancer Centre
Appointed Sep 2023

Adam Horsburgh

Chief Executive Officer
Austin Health
Appointed Mar 2017

Prof Andrew Stripp

Chief Executive
Monash Health
Appointed May 2016

Dr Dan Grant

Independent Director
Appointed Dec 2021
Retired from Board Mar 2023

Finance, Audit and Risk Committee

Adam Horsburgh (Chair)

Chief Executive Officer
Austin Health
Appointed May 2021

Lucy Franzmann

Chief Financial Officer
Victoria University
Appointed May 2021

Joh Kirby

Head – Governance, Risk and Compliance
Walter and Eliza Hall Institute
Appointed May 2021

Deb Sullivan

Chief Financial Officer
Murdoch Children's Research Institute
Appointed May 2022

Business Development and Commercialisation Committee:

Dr Dan Grant

Independent Chair
Appointed Dec 2021
Retired from Committee Mar 2023

Lynne Cobiac

Director (Acting) Health and Biosecurity
CSIRO
Appointed May 2023

Dr Angus Henderson

Director, Enterprise Strategy and Development
Monash Health
Appointed May 2022

Dr Anne-Laure Puaux

Head – Biotechnology and Commercialisation
Walter and Eliza Hall Institute
Appointed May 2022

Dr Janet Fox

Business Development Manager
CSIRO
Appointed May 2022

Joe Baini

Chief Executive Officer
Australian Genome Research Facility
Appointed May 2022

“The brilliance of Melbourne Genomics was that it started with the actual institutions. It started with the CEOs. Right from the very beginning there was a sense of partnership and a sense of buy-in at the very highest level and it made all the difference in terms of the way it was accepted, and the way that people were interested in it.”

- Stakeholder (quote provided for formative evaluation)

Community voices

Patient and community perspectives are critical to the safe provision of genomic medicine.

Community Advisory Group (CAG)

Since its formation in 2014, the CAG has advised on every Melbourne Genomics project – helping to determine what kind of engagement is needed, and with whom.

The CAG met four times in 2023. They informed the development of project-based consumer panels, contributed to public consultations, and provided guidance on inclusion and equity.

Kellie-Ann Jolly (Chair)

Appointed Aug 2022

Louisa Di Pietro

Appointed Jan 2014
Retired from CAG Aug 2023

Stacey Ong

Appointed Aug 2022

Heather Renton

Appointed Jan 2014

Margaret Sahhar AM

Appointed Jan 2014

Spase Veljanovski

Appointed May 2022

Dr Janney Wale

Appointed Jan 2014

Christine Walker

Appointed Jan 2014

Sue Emery

Appointed Nov 2023

Maya Pinn

Appointed Nov 2023

[Visit our website for member profiles](#)

Consumer panels

On advice from the CAG, we formed four new consumer panels – bringing together people with lived experience to inform our clinical projects.

Cancer Statewide project

Bonney Corbin
Jo Cockwill
Victoria Sharp

Dementia Clinical Change project

Paul Flavel
Isabelle Burke
Felicity Peel
Mardi S
Sally M

Nephrology Clinical Change project

Robert Gordos
Hannah Passmore
Mitch Passmore
Nelly Counihan
Tom Greaterox
Lisa Bell

Transplant Clinical Change project

Lorien Kaye
Andrew Chapman
Albert Fenech
Susan Wilson
Chris D
Nada F

Paediatrics Statewide project

Heather Renton
Hollie Feller
Maya Pinn

Some consumers opted not to be identified. We respect their privacy and are grateful for their many contributions.

ALIGN (Australian Alliance for Indigenous Genomics)

Led by Indigenous researchers and health professionals, ALIGN seeks to ensure that Aboriginal and Torres Strait Islander peoples benefit explicitly and equitably from genomics.

This year, Melbourne Genomics was proud to provide administrative support to the Victorian node of ALIGN and its governance committee.

Indigenous Governance Committee

Shawana Andrews

The University of Melbourne

Shayne Bellingham

LIME Network and The University of Melbourne

Kate Brocker

First Peoples' Health and Wellbeing

Jill Gallagher

VACCHO (Jim O'Shea, proxy)

John Gorton

Goolum Goolum Aboriginal Co-operative

Tala Mitchell

SING and Deakin University

Paul Patten

Community Member

Implementation Fellowships

Implementation Fellowships support people to progress the use of genomics in healthcare.



Antibiotic resistance

Dr **Stefano Giulieri**, from the University of Melbourne and The Royal Melbourne Hospital, is combining advanced microbiological testing and bacterial genomics to better understand, detect and help doctors respond to antibiotic failure.



Cardiology

Genetic counsellor **Heather Chalinor** is working with the Austin Health cardiology team to help them incorporate genetic testing into their cardiac clinics.



Cloud computing

Labs often use Secure File Transfer Protocol (SFTP) to move large genomic data files from one computer program to the next. Dr **Lesley Gray** at AGRF is testing Genomical's cloud-based platform as an alternative, enabling data to stay within the cloud for each step of analysis.



Deafness

Medicare now funds genomic testing for the causes of childhood deafness. While laboratories prepare to provide testing, geneticist **Dr Lilian Downie** is helping doctors get ready to offer this test to their patients.



Dementia care

Neurologist **Dr Oneil Bhalala** at The Royal Melbourne Hospital is exploring the role of 'omic technologies for diagnosis and prediction in older-age dementia patients. He is particularly interested in the use of 'polygenic risk scores' – which were developed primarily based on European ancestry – and whether they need to be modified to reflect the ethnic diversity of Australia.



Diagnostic pathology

In November 2023, Medicare announced funding for a gene test for blood cancers. A/Prof **Piers Blombery** is working to ensuring Peter Mac's molecular haematology laboratory can scale up and meet anticipated demand.



Dermatology

Dermatologist **Dr Lauren Anderson** is building her expertise in using genomics to inform the diagnosis and care of patients with genetic skin conditions at The Royal Melbourne Hospital.



Infectious respiratory disease

Superbugs (antibiotic-resistant microbes) can be especially harmful to immunocompromised patients. A/Prof **Norelle Sherry**, from the University of Melbourne's Centre for Pathogen Genomics, is testing the use of a metagenomics panel that can analyse the genetic makeup of a wide range of bacteria, viruses and fungi within patient samples.



Informed consent

Patients consent to genomic testing by signing forms, which are often saved as PDFs or images together with multiple other documents. Dr **Simon Sadedin's** team at the Murdoch Children's Research Institute will trial optical character recognition technology to extract text-based information from the images, followed by natural language processing methods to identify what each patient has consented to.



Inherited conditions

Dr **Amit Kumar** at Monash Health is preparing a whole exome sequencing workflow, using genomic sequencing data received from the Australian Genome Research Facility (AGRF). By outsourcing the genomic sequencing stage of the process, labs can avoid having to buy expensive DNA sequencers, and focus instead on analysing the genomic data.



Haematology

Haematologists **Dr Lucy Fox** and **Dr Ashvind Prabakaran** have established a genetic haematology clinic at Austin Health to ensure patients receive optimal care for blood cancers. This includes identifying gene changes that can cause blood cancers within a family, and screening relatives who wish to donate bone marrow for a transplant.



Maternal and foetal medicine

Obstetrician **Dr Ted Springhall** is bringing genomics into maternal and foetal medicine at Monash Health, to ensure patients who've had abnormal ultrasound findings get access to appropriate genomic testing and counselling.



Paediatric rheumatology

Paediatric rheumatologist **Dr Georgina Tiller** is reviewing the different kinds of genetic and genomic tests provided to patients at The Royal Children's Hospital. This will inform identify which tests are most useful for the different patients seen in paediatric rheumatology, pathways for ordering these and systems to track testing.



Pharmacogenomics

Pharmacist **Ben Lee** is developing pharmacogenomic education modules for a wide range of healthcare professionals who prescribe and provide medications for patients.



Reporting and analysis

CSIRO has developed sBeacon, a cloud-based tool that enabled researchers to share and analyse genomic variants, to gain medical insights without compromising patient privacy. Dr **Denis Bauer's** team is now testing the reporting capabilities of sBeacon within the Genomical software platform.

Clinical project teams

Melbourne Genomics projects are led and implemented by clinicians, scientists and researchers from across the Alliance.

These experts will continue to take genomics forward long after the projects are complete.

Controlling Superbugs project

Project lead

Prof Lindsay Grayson, Austin Health

Project coordinator

Dr Dorothy Ling, Austin Health

Project team

A/Prof Jason Kwong, Austin Health

A/Prof Norelle Sherry, The University of Melbourne/The Doherty Institute

Prof Benjamin Howden, The University of Melbourne/The Doherty Institute

Project site team

Prof Rinaldo Bellomo, Austin Health

Jordana Flude, Austin Health

Elizabeth Grabsch, Austin Health

Dr Natasha Holmes, Austin Health

Leanne Houston, Austin Health

Dr Marcel Leroi, Austin Health

Jolian Poliss, Austin Health

Kartik Kishore, Austin Health

Kathryn Cisera, Monash Health

A/Prof Maryza Graham, Monash Health

A/Prof Tony Korman, Monash Health

Despina Kotsanas, Monash Health

Stella Snibson, Monash Health

Prof Rhonda Stuart, Monash Health

Hayley Page, Peter MacCallum Cancer Centre

Prof Monica Slavin, Peter MacCallum Cancer Centre

Rachel Woolstencroft, Peter MacCallum Cancer Centre

A/Prof Leon Worth, Peter MacCallum Cancer Centre

A/Prof Andrew Daley, The Royal Children's Hospital

Gena Gonis, The Royal Children's Hospital

Sophie Kyriakou, The Royal Children's Hospital

Dr Lai-Yang Lee, The Royal Children's Hospital

Dr Katherine Bond, The Royal Melbourne Hospital

Lizia Lai, The Royal Melbourne Hospital

Lisa Marks, The Royal Melbourne Hospital

A/Prof Caroline Marshall, The Royal Melbourne Hospital

Rica Tamala, The Royal Melbourne Hospital

Cancer Statewide project

Project lead

Prof Jayesh Desai, Peter MacCallum Cancer Centre

Expert site leads

Dr Kortnye Smith and Dr Lavinia Tan, Peter MacCallum Cancer Centre

Implementation science lead

A/Prof Laura Forrest, Peter MacCallum Cancer Centre

Laboratory lead

Prof Stephen Fox, Peter MacCallum Cancer Centre

Project coordinator

Christine Goulter, Peter MacCallum Cancer Centre

Research assistant

Kim An, Peter MacCallum Cancer Centre

Project team

Denis Cleven, Peter MacCallum Cancer Centre

Prof Stephen Fox, Peter MacCallum Cancer Centre

Christine Goulter, Peter MacCallum Cancer Centre

Dr Chelsee Hewitt, Peter MacCallum Cancer Centre

Shravan Yellenki, Peter MacCallum Cancer Centre

Dr Huiling Xu, Peter MacCallum Cancer Centre

Dr Michelle Tew, The University of Melbourne

A/Prof Natalie Taylor, UNSW School of Population Health

Project site leads and local superusers

Prof Gary Richardson, Cabrini Health

Dr Arvind Sahu, Goulburn Valley Health

Dr Sharad Sharma, Grampians Health

Dr Sachin Joshi, Latrobe Regional Health

Dr Daphne Day, Monash Health

Dr Belinda Lee, Northern Health

Project site investigators

Dr Prachi Bhave, Cabrini Health

Dr David Cheng, Goulburn Valley Health

Dr Myron Klevansky, Goulburn Valley Health

Dr Nikhl Pande, Goulburn Valley Health

Dr Javier Torres, Goulburn Valley Health

Dr Samantha Dean, Grampians Health

Dr James Ridgwell, Grampians Health

Dr Daniel Stout, Grampians Health

Dr Hieu Chau, Latrobe Regional Health

Dr Alison Hiong, Latrobe Regional Health

Dr Jason Qin, Latrobe Regional Health

Dr Bhavini Shah, Latrobe Regional Health

Dr Amy Body, Monash Health

Dr Sophia Frentzas, Monash Health

Dr JB Kong, Monash Health

Dr Jeremy Neeman, Monash Health

Dr Jessica Nommo, Monash Health

Dr Christian Orłowski, Monash Health

Dr Sammy Shaya, Monash Health

Dr Edbert Wong, Monash Health

Dr Azim Jalali, Northern Health

Dr Jennifer Mooi, Northern Health

Dr Abdelaziz Sammour, Northern Health

Dr Richard To, Northern Health

Paediatrics Statewide project

Project lead

A/Prof Belinda McClaren

Project Steering Group

This group provided advice on how best to address the challenges of increasing uptake of funded exome testing for childhood syndromes by paediatricians.

Hollie Feller, Genetic Support Network of Victoria

Dr Emma Weisz, Melbourne Genomics Health Alliance

A/Prof Michael Fahey, Monash Health

Dr Luisa Clucas, The Royal Children's Hospital

Heather Renton, Founder and former CEO of Syndromes Without A Name (SWAN)

Dr Natasha Brown, Victorian Clinical Genetics Services

Justine Elliott, Victorian Clinical Genetics Services

Working Group

Dr Andrew Fennel, Monash Health

Anita Gorrie, Monash Health

Justine Elliott, Victorian Clinical Genetics Services

Ivan Macciocca, Victorian Clinical Genetics Services

Dr Alison Yeung, Victorian Clinical Genetics Services

The following people contributed to implementation of this project in 2023.

Dr Angus Henderson, Monash Health

Prof Mike South, The Royal Children's Hospital

Justine Elliott, Victorian Clinical Genetics Services

Ivan Macciocca, Victorian Clinical Genetics Services

Dr Meg Wall, Victorian Clinical Genetics Services

Hospital Implementation Project

Project lead

A/Prof Cate Kelly

Research team

Dr Trang Do, Murdoch Children's Research Institute

A/Prof Belinda McClaren, Murdoch Children's Research Institute

Hospital Implementation Reference Group

This group provided advice on how best to help hospitals address the challenges of bringing genomics into routine care.

Rachel Meehan, Austin Health (retired from HIRG February 2023)

Charlie MacArthur, Austin Health

Dr Jason Goh, Monash Health

Prof John Seymour AM, Peter MacCallum Cancer Centre

Dr David Speakman, Peter MacCallum Cancer Centre (retired from HIRG June 2023)

A/Prof Tom Connell, The Royal Children's Hospital

Dr Fergus Kerr, The Royal Melbourne Hospital

Two working groups, formed in 2023, contributed to this project.

Professional Governance of Genomic Medicine in Hospitals

Dr Lucy Fox, Austin Health

Gina McLachlan, Austin Health

Dr Junyi Shi, Goulburn Valley Health

Dr Jason Goh, Monash Health

Dr Kushani Jayasinghe, Monash Health

Prof Beena Kumar, Monash Health

A/Prof Tom Connell, The Royal Children's Hospital

Dr Aamira Huq, The Royal Melbourne Hospital

Dr Fergus Kerr, The Royal Melbourne Hospital

Prof Paul James, The Royal Melbourne Hospital and Peter MacCallum Cancer Centre

Elly Lynch, Victorian Clinical Genetics Services

Measuring Quality of Genomic Medicine Care

Prof Kerry Ireland-Jenkin, Austin Health

Charlie McArthur, Austin Health

Prof Gary Richardson OAM, Cabrini Hospital

Monica Ferrie, Genetic Support Network of Victoria

Dr Janney Wale, Melbourne Genomics Community Advisory Group

Michaela Cormack, Monash Health

Dr Kushani Jayasinghe, Monash Health

Dr Gráinne Butler, Murdoch Children's Research Institute

A/Prof Cathy Quinlan, The Royal Children's Hospital

Dr Tim Fazio, The Royal Melbourne Hospital

Dr Aamira Huq, The Royal Melbourne Hospital

Bryony Thompson, The Royal Melbourne Hospital

Prof Paul James, The Royal Melbourne Hospital and Peter MacCallum Cancer Centre

A/Prof Chris Schilling, The University of Melbourne

Clinical Change projects

Research team

Dr Belinda McClaren, Murdoch Children's Research Institute

Dr Trang Do, Murdoch Children's Research Institute

Transplant

Project lead

Prof Paul James, The Royal Melbourne Hospital

Project coordinator

Dr Lokman Pang, The Royal Melbourne Hospital

Project site teams

Linda Ciciarelli, Austin Health

Gina McLachlan, Austin Health

Dr Karl Vaz, Austin Health

Christy Atkinson, The Royal Melbourne Hospital

Susan Fisher, The Royal Melbourne Hospital

Dr Stephanie Kuo, The Royal Melbourne Hospital

Sangavi Sivagnanasundram, The Royal Melbourne Hospital

Dr Bryony Thompson, The Royal Melbourne Hospital

Collaborators

Kent Garrett, Austin Health

Graham Starkey, Austin Health

Tim Tran, Austin Health

Joe Bains, Australian Genome Research Facility

Dr Steven Bentley, Australian Genome Research Facility

Dr Kenneth Chan, Australian Genome Research Facility

Melanie O'Keefe, Australian Genome Research Facility

Jacqueline Montgomery, Australian Genome Research Facility

Dr Cath Moore, Australian Genome Research Facility

Matthew Tinning, Australian Genome Research Facility

Dr Michael Christie, The Royal Melbourne Hospital

A/Prof Peter Hughes, The Royal Melbourne Hospital

Prof Ingrid Winship, The Royal Melbourne Hospital

Nephrology

Project lead

Dr Kushani Jayasinghe, Monash Health

Project coordinator

Briannah Miles, Monash Health

Project site team

Jen Bakker, Austin Health
Anna Leaver, Austin Health
Dr Mia Leung, Austin Health
Giulia Valente, Austin Health
Paula Bussa, Monash Health
Dr Ben Lazarus, Monash Health
Kathryn Visser, Monash Health
Dr Kenneth Xie, Monash Health
Brendan Cusack, The Royal Children's Hospital
Yoni Elbaum, The Royal Children's Hospital
Dr Cathy Quinlan, The Royal Children's Hospital
Ella Wilkins, The Royal Children's Hospital
Dr Mandy Law, The Royal Melbourne Hospital
Andrea Ward, The Royal Melbourne Hospital
Jack Wheeler, The Royal Melbourne Hospital
Prof Zornitza Stark, Victorian Clinical Genetics Services

Collaborators

Dr Simon Bodek, Austin Health
Dr Ainsley Campbell, Austin Health
A/Prof John Whitlam, Austin Health
Prof Peter Kerr, Monash Health
Dr Matthew Regan, Monash Health
Dr Russell Gear, The Royal Melbourne Hospital
Prof Paul James, The Royal Melbourne Hospital
Dr Kathy Nicholls, The Royal Melbourne Hospital
Dr Bryony Thompson, The Royal Melbourne Hospital
A/Prof Stephanie Best, University of Melbourne
Dr Lin Cheng, University of Melbourne
A/Prof Sebastian Lunke, Victorian Clinical Genetics Service

Dementia

Project lead

Dr Aamira Huq, The Royal Melbourne Hospital

Co-investigator

A/Prof Adrienne Sexton, The Royal Melbourne Hospital

Project coordinator

Charlotte Webster, The Royal Melbourne Hospital

Project site team

Ashley Crook, Austin Health
Nikki Gelfand, Monash Health
Dr Caitriona Monahan, The Royal Melbourne Hospital
Dr Alexandra Waxmann, The Royal Melbourne Hospital
Kirsty West, The Royal Melbourne Hospital

Collaborators

Dr Ainsley Campbell, Austin Health
Prof Martin Delatycki, Austin Health
A/Prof Michael Woodward, Austin Health
Dr Susan Mathers, Calvary Health
Prof Amy Brodtman, Eastern Health
Dr Michael Fahey, Monash Health
Dr Matthew Hunter, Monash Health
Prof Henry Ma, Monash Health
Prof Dennis Velakoulis, The Royal Melbourne Hospital
Prof Ingrid Winship, The Royal Melbourne Hospital
Dr Christine Wools, The Royal Melbourne Hospital
Prof Melanie Bahlo, WEHI

Equity and Access – Data project

Project lead

Prof Angeline Ferdinand, The University of Melbourne

Project officer

Nadine Phillips, The University of Melbourne

Indigenous Genomics Health Equity project

Project lead

Sara Alden, VACCHO

Project team

Olivia Payne, VACCHO
Jim O'Shea, VACCHO
Louise Lyons, ALIGN

Health Economics

Project lead

A/Prof Chris Schilling, The University of Melbourne

Research assistant

Florencia Putri Sjaaf, The University of Melbourne



Expert educators

An expert advisory group contributed to our education strategy.

Michelle Barrett

VCCC Alliance

A/Prof Femke Buisman-Pijlman

Melbourne School of Professional and Continuing Education

Prof John Christodoulou

The University of Melbourne

Prof Stephen Fox

Peter MacCallum Cancer Centre

Prof Amy Gray

The Royal Children's Hospital

Prof Steve Trumble

The University of Melbourne

Prof Jenny Weller-Newton

The University of Melbourne

Multiple clinicians, scientists and other specialists helped design workshops or shared their expertise as facilitators.

Austin Health

Dr Simon Bodek
Heather Chalinor

Bendigo Health

Justin Hargreaves

Cabrini Family Cancer Clinic

A/Prof Yoland Anthill

Calvary Mater Newcastle

Gillian Blanchard

Cancer Nurses Association of Australia

Jemma Still

Eastern Health

Dr Patrick Carney

Genetic Support Network of Victoria

Monica Ferrie

Monash Health

Dr Kushani Jayasinghe
Gillian Kruss

Monash University

Dr Jane Tiller

Murdoch Children's Research Institute

Dr Yan Tu

MyDNA

Sam Mostafa

NSW Health Pathology

Corrina Cliffe

Peter MacCallum Cancer Centre

Dr Victoria Bashay

Dr Michael KC Lee

Dr Chris McEvoy

Dr Georgina Ryland

Dr Kortnye Smith

Dr Lavinia Tan

A/Prof Alison Trainer

Dr Huiling Xu

SWAN Australia

Heather Renton

Tasmanian Clinical Genetics Service

Dr Mathew Wallis

The Royal Children's Hospital

Dr Kanita Bhatia

Professor Lynn Gillam

Dr Katherine Howell

Professor Rick Leventer

Dr Dong Anh Khuong Quang

Dr Nicholas Sanders

A/Prof Valerie Sung

The Royal Melbourne Hospital

Dr Lauren Akesson

Dr Mark Cleghorn

Dr Russell Gear

Prof Paul James

Joshua Schultz

A/Prof Adrienne Sexton

Dr Bryony Thompson

Dr Christine Wools

The University of Melbourne

Prof Sean Grimmond

Dr Richard Rebello

Dr Sibel Saya

Dr Joep Vissers

Dr Layla Zhu

Victorian Clinical Genetics Services

Sam Ayres

Dr Sunita Biswas

Dr Natasha Brown

Dr Chloe Cunningham

Lisette Curnow

Prof Martin Delatycki

Dr Michelle De Silva

Dr Lillian Downie

Justine Elliot

Lyndon Gallacher

Manny Jacobs

A/Prof Sebastian Lunke

Elly Lynch

Dr Krithika Murali

Prof Zornitza Stark

Prof Tiong Tan

Prof Sue White

Ella Wilkin

Networking events and webinars helped students and professionals alike to see the possibilities of a career in genomics. The following experts co-hosted or helped facilitate these events.

A/Prof Bernie Pope Australian BioCommons

Dr Gad Abraham CSL Research

Dr Bo Xu Illumina

Dr Khalid Mahmood Melbourne Bioinformatics

Dr Alicia Arnott Peter Doherty Institute for Infection and Immunity

Dr Ash Porter, Peter Doherty Institute for Infection and Immunity

Dr Dineika Chandrananda, Peter MacCallum Cancer Centre

Prof Kim-Anh Lê Cao, The University of Melbourne

A/Prof Agus Salim, The University of Melbourne

Dr Noel Faux, The University of Melbourne

Dr Benjamin Goudey, The University of Melbourne

Kirsten Doert-Eccles, The University of Melbourne

Dr Justin Bedó, WEHI

Data and technology experts

Genomics, bioinformatics and medical science experts contributed to the ongoing development of the Genomical software platform, as well as to data governance, privacy and security projects.

Alfred Health

Jane Lin
Adam Ivey
Prof Andrew Perkins

Austin Health

Dr Chris Hogan
A/Prof Kerryn Ireland-Jenkins
Dr Jonathon Clark
Dr Rishu Agarwal

Australian Genome Research Facility

Dr Lesley Gray
Dr Matthew Tinning

CSIRO

Dr Denis Bauer
Dr Yatish Jain
Dr Anuradha Wickramarachchi

Monash Health

Dr Paul Yeh
Dr Ravikiran Vedururu
Dr Amit Kumar

The Royal Melbourne Hospital

Dr Tim Fazio
Dr Bryony Thompson
Adam Boulton
George Cozaris
Dr Giles Kelsey

Victorian Clinical Genetics Services

Dr Sebastian Lunke
Dr Simon Sadedin
Revi Rosenberg
Ashil Davawala

Innovation and Prioritisation Committee

Dr Cath Moore, Australian Genome Research Facility
Dr David Hansen, CSIRO
Dr Angus Henderson, Monash Health
Revital Rosenburg, Murdoch Children's Research Institute
Dr Tim Fazio, The Royal Melbourne Hospital
Prof Paul James, The Royal Melbourne Hospital
Dr Vineesh Khanna, Victorian Department of Health

Project Control Board

Dr David Hansen, CSIRO,
Simon Cowley, Victorian Department of Health
George Cozaris, The Royal Melbourne Hospital

Program team

Each Melbourne Genomics project is supported by a team with expertise ranging from research, program design and evaluation to project management, education, informatics, software development, communications and community engagement.

The following people were part of the program team in 2023.

Prof Clara Gaff
Katie Arkell
Danielle Ariti
Manav Arora
Tim Bakker
Daniela Bodemer
Naomi Burke
Amy Clarke
David Cloake
Dr Luisa Clucas
Vicki Crowley
Zayne D'Crus
Irene Dinatale
Jaitika Duggal

Lauren Eldershaw
Caroline Foster
Dr Tim Hastings
Chriselle Hickerton
Kristina Hood
Jessica Ince
Navdeep Kaur
Maira Kentwell
Michelle Kleynhans
Douglas Liddicoat
Dr Fran Maher
Dr Melissa Martyn
Daniel Mason
Matt Nielsen

Dr Amy Nisselle
James Oakes
Kitty O'Brien
Amelia Rahardja
Keeley Reade
Peter Scott
Nisha Subramanian
Rigan Tytherleigh
Dr Natalie Thorne
Jordan Ung
Dr Emma Weisz
Caroline Zhang

The Melbourne Genomics staff team at the '10 Years of Genomics' event in November 2023.
Photo: Melbourne Genomics/Thomas Elia



Where you saw us in 2023

Date	Details	Presenter
24/02/2023	2023 International Congress of Human Genetics , Invited session: Ensuring Best Practice in Genomics Education and Evaluation: The Australian Genomics Workforce and Education Research Program	Dr Amy Nisselle
24/02/2023	2023 International Congress of Human Genetics , A staged approach to developing the genomics workforce while implementing genomic medicine	Dr Amy Nisselle
22/03/2023	InGeNA Webinar , Invited presentation: Addressing NAGIM Implementations	Dr Natalie Thorne
4/04/2023	WEHI Bioinformatics Seminars , How to make a shotgun marriage last a lifetime	Dr Natalie Thorne
14/04/2023	Victorian Government and Roche Diagnostics MedTech Showcase , GenoVic	Michelle Kleynhans
20/04/2023	Australian Genomics' CDR Network Meeting , Invited presentation: Everything, Everywhere, All at Once (Almost): About Melbourne Genomics	Prof Clara Gaff
28/04/2023	Customer Vulnerability Symposium , How to make strength-based storytelling an everyday practice	Zayne D'Crus
12/05/2023	Monash Children's Paediatric Update , paediatricgenomics.org.au all-in-one resource hub to support paediatricians with Medicare-funded genomic testing	Dr Emma Weisz
13/05/2023	Melbourne Children's Paediatric Update , paediatricgenomics.org.au all-in-one resource hub to support paediatricians with Medicare-funded genomic testing	Dr Emma Weisz
20/05/2023	Hong Kong Genome Institute's Embracing the era of genomic medicine symposium , Invited presentation: Preparing clinicians for the genomic medicine era: forntline workforce training and education	Prof Clara Gaff
6/06/2023	Digital Health Festival 2023 , Understanding realities, exceeding expectations	Dr Natalie Thorne
8/07/2023	MedInfo 2023 , Panel on Genomic Data Sharing: Balancing Connectivity Insight and Consumer Control	Keeley Reade, Dr Natalie Thorne, Dr Tim Fazio, Colleen Brooks and Gregg Pratt
10/07/2023	MedInfo 2023 , Insight from the proven implementation of a digital health platform for genomics	Dr Natalie Thorne
15/07/2023	UK Implementation Science Conference , Using logic models to advance the implementation of complex genomics sequencing within a complex care pathway	Joey Elias

15/07/2023	2023 International Congress of Genetics , Satellite Meeting/ Genomics Update: New technologies and clinical cases - Has genomics met its promise?	Prof Lynn Gillam, Dr Alison Archibald, Bonney Corbin, Prof Clara Gaff, A/Prof Sebastian Lunke, Dr Jane Tiller, A/Prof Alison Trainer
19/07/2023	2023 International Congress of Genetics/HGSA Annual Scientific Meeting , Invited presentation (Sutherland Lecture): Accelerating rare disease diagnosis	Prof Zornitza Stark
20/07/2023	2023 International Congress of Genetics , Industry Breakfast: Missed connections, why genomics and digital health aren't getting along	Dr Natalie Thorne
27/07/2023	MCRI REDCap User Group Forum , Accesibility and REDCap Surveys	Rigan Tytherleigh
15/08/2023	Leaders Within presented by SAHMRI and CALHN , The DNA of leadership in the era of digital disruption	Dr Natalie Thorne
21/09/2023	WHO Western Pacific Regional Office Manila , "Third Meeting of the Western Pacific Region Emerging Molecular Pathogen Characterization Technologies (EMPaCT) Surveillance Network, 21-22 September 2023, Manila, Philippines"	A/ Prof Norelle Sherry
8/10/2023	Evidence and Implementation Summit Satellite Workshop , Understanding the changing roles of genetic counsellors in facilitating the wider use of genomics in patient care in Australia	Dr Trang Do
9/10/2023	Evidence and Implementation Summit , Designing interventions to support use of complex genomic profiling in advanced care	A/Prof Belinda McClaren
10/10/2023	Evidence and Implementation Summit , Designing interventions to support use of complex genomic profiling in advanced cancer care	Dr Melissa Martyn
10/10/2023	Evidence and Implementation Summit , Implementing genomics: lessons learnt from the Melbourne Genomics Health Alliance 2014-2024	Dr Melissa Martyn
13/10/2023	MCRI Postdoc Symposium , Evaluating co-designed interventions supporting paediatricians to order funded genomic tests: a website, 'ask-a-genetics-expert' contact service and awareness raising activities	A/Prof Belinda McClaren
9/11/2023	Wild Health Sydney Summit - Australia vs the World , Panel on Israel's amazing population data paradigm	Dr Natalie Thorne
9/11/2023	Wild Health Sydney Summit - Australia vs the World , Panel on Estonia's astonishing system transformation	Dr Natalie Thorne
18/11/2023	Australasian Society of Genetic Counsellors Special Interest Group , Evaluating co-designed interventions supporting paediatricians to order funded genomic tests: a website, 'ask-a-genetics-expert' contact service and awareness raising activities	A/Prof Belinda McClaren
18/11/2023	Australasian Society of Genetic Counsellors Special Interest Group , Becoming Agents for Genomic Change: Genetic Counsellors' Views of Patient Care and Implementation Influences when Genomics is Mainstreamed	Dr Trang Do
27/11/2023	International Genetics Education and Training Summit , Evaluating genetics/genomics education initiatives	Dr Amy Nisselle
27/11/2023	International Genetics Education and Training Summit , The Australian perspective: A toolkit for genomics educators	Prof Clara Gaff
28/11/2023	PHG Foundation, Cambridge, UK Preparing clinicians for the genomic medicine era	Prof Clara Gaff
4/12/2023	Symposium on Bioinformatics Engineering in Industry by ABACBS , Mind. Blown. (What a bioinformatician learned about doing business). Dr Natalie Thorne on why building a great product is just the first tiny step.	Dr Natalie Thorne

Peer-reviewed publications

“Genomics education for medical specialists: case-based specialty workshops and blended learning” Fran Maher, Amy Nisselle, Elly Lynch, Melissa Martyn, Rigan Tytherleigh, Taryn Charles, Clara Gaff, *Journal of Translational Genetics and Genomics*. 7, no.2: 94-109. <http://dx.doi.org/10.20517/jtgg.2023.04>

“Offering and returning secondary findings in the context of exome sequencing for hearing loss: Clinicians’ views and experiences”, Lauren Notini, Clara Gaff, Julian Savulescu and Danya F Vears, *AJOB Empir Bioeth*. 2023 Apr-Jun;14(2):74-83. doi: [10.1080/23294515.2022.2160507](https://doi.org/10.1080/23294515.2022.2160507).

“What matters to parents? A scoping review of parents’ service experiences and needs regarding genetic testing for rare diseases”, Erin Crellin, Melissa Martyn, Belinda McClaren and Clara Gaff, *European Journal of Human Genetics* 31, 869–878 (2023). <https://doi.org/10.1038/s41431-023-01376-y>

Related publication

“Two-step offer and return of multiple types of additional genomic findings to families after ultrarapid trio genomic testing in the acute care setting: a study protocol”, Sophie E Bouffler, Ling Lee, Fiona Lynch, Melissa Martyn, Elly Lynch, Ivan Macciocca, Lisette Curnow, Giulia McCorkell, Sebastian Lunke, Belinda Chong, Justine E Marum, Martin Delatycki, Lilian Downie, Ilias Goranitis, Danya F Vears, Stephanie Best, Marc Clausen, Yvonne Bombard, Zornitza Stark and Clara Gaff. *BMJ Open* 2023;13:e072999. doi:[10.1136/bmjopen-2023-072999](https://doi.org/10.1136/bmjopen-2023-072999)

Operations

Our focus in 2023 was to work with the Alliance member organisations and the Victorian Government to establish a new company to manage the Genomical® platform.

Funding and governance

The program was administered by the Victorian Department of Health in 2023.

The Department stewarded the complex approval process through government to enable the establishment of the new company, Transcendomics.

The Business Development & Commercialisation Committee has provided expert oversight of alliance business development activities. This committee concluded its work in 2023 having ensured the sustainability of program assets of enduring value.

Business Development

The expertise of the Alliance Board and its committees provided strategic guidance through each phase of the planning to commercialise the Genomical platform.

The governance and structure of the new company, jointly owned by the member organisations and the Victorian Government, was approved in 2023.

A business case was developed to determine how genomic education developed by the Alliance could be sustainable beyond 2025. The business case was informed by the Licensing model for education and training that was expanded in 2023. The recommended approach will be implemented in 2024.

Engagement

Community engagement activities have enhanced the integration of lived experience into program delivery. Guided by the Melbourne Genomics Community Advisory Group, six consumer panels provided input into clinical projects. Meanwhile, a project commenced with VACCHO to inform equity in genomic healthcare for Aboriginal and Torres Strait Islander people in Victoria.

Immersion fellowships and implementation projects were funded by the Alliance in each of the member organisations to build capability and address challenges and barriers to the implementation of genomics in health care.

Program finalisation

Planning has commenced for the staged finalisation of the program in 2025, including financial acquittal and ongoing data management.

Financial statements

for the year ended 31 December 2023

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Statement of profit or loss and other comprehensive income

for the year ended 31 December 2023

	Note	2023 \$'000	2022 \$'000
Operating revenue			
Grant revenue		10,690	6,588
Member contributions		2,500	2,500
Interest income		602	199
Sundry income		11	26
Total operating revenue		13,803	9,313
Operating expenditure			
Staff costs	2(b)	3,541	2,832
Consumable supplies		24	48
Consultancy		8,872	5,866
Other expenses		1,366	567
Total operating expenditure		13,803	9,313
Surplus from operations		-	-

The financial statements are to be read in conjunction with the notes to and forming part of the financial statements.

Statement of financial position

for the year ended 31 December 2023

	Note	2023 \$'000	2022 \$'000
Assets			
Current assets			
Cash and bank balances	6(a)	8,457	
Term deposits	6(a)	4,000	
Other financial assets		-	
Trade and other receivables	3	3,570	
Total current assets		16,027	
Total assets		16,027	
Liabilities			
Current liabilities			
Trade and other payables	4	1,356	
Unearned grant income	5	14,671	
Total current liabilities		16,027	
Total liabilities		16,027	
Net assets		-	
Total funds		-	

The financial statements are to be read in conjunction with the notes to and forming part of the financial statements.

Statement of cash flows

for the year ended 31 December 2023

	Note	2023 \$'000	2022 \$'000
Cash flows from operating activities			
Receipts from sundry income		12	26
Receipt from granting bodies		4,915	14,745
Receipt from member contributions		3,750	2,092
Payments to suppliers and employees		(15,281)	(7,799)
Interest received		639	279
Net cash (used in)/generated by operating activities	6(b)	(5,965)	9,343
Cash flows from investing activities			
Receipts from (investments) in financial assets		6,000	(6,000)
Net cash generated by/(used in) investing activities		6,000	(6,000)
Net cash used in financing activities			
		-	-
Net increase in cash and held		35	3,343
Cash and cash equivalents at the beginning of the period		12,422	9,079
Cash and cash equivalents at the end of the period	6(a)	12,457	12,422

The financial statements are to be read in conjunction with the notes to and forming part of the financial statements.

Statement of changes in equity

for the year ended 31 December 2023

	Total Funds 31 December 2023 \$'000
Balance at 1 January 2022	-
Operating surplus for the period	-
Balance as at 31 December 2022	-
Operating surplus for the period	-
Balance at 31 December 2023	-

The financial statements are to be read in conjunction with the notes to and forming part of the financial statements.

Notes to the financial statements

for the year ended 31 December 2023

1. Statement of significant accounting policies

The entity is an unincorporated Alliance and has prepared the financial statements on the basis that it is a non-reporting entity because there are no users dependent on a general-purpose financial report. The financial report is therefore a special-purpose financial report that has been prepared in order to meet the requirements of the unincorporated alliance.

Basis of preparation

The financial report has been prepared on the basis of historical cost except for the revaluation of certain non-current assets and financial instruments. Cost is based on the fair values of consideration given in exchange for assets.

The financial statements have been prepared in accordance with the recognition and measurement requirements specified by all Australian Accounting Standards and Interpretations, and the disclosure requirement of Accounting standards:

- AASB 101 Presentation of Financial Statements
- AASB 107 Statement of Cash Flows
- AASB 108 Accounting Policies Changes in Accounting Estimates and Errors
- AASB 1048 Interpretation and Application of Standards
- AASB 1054 Australian Additional Disclosures,

Accounting policies are selected and applied in a manner which ensures that the resulting financial information satisfies the concepts of relevance and reliability, thereby ensuring that the substance of the underlying transactions or other events is reported.

A funding agreement between the Alliance and the State of Victoria was signed on 9th June

2021. The funding agreement of \$35million is for the contribution to the continuation of the Melbourne Genomics Health Alliance. The current executed collaboration agreement between the Alliance members has been extended until 31 December 2025 after a new collaboration agreement was fully signed on 11th February 2022. The financial statements are presented on a going concern basis.

The financial statements are presented in Australian Dollars.

The financial statements include all the activities of Phase 3 of Melbourne Genomics Health Alliance.

Principal address of the Alliance is:

1G Royal Parade
Parkville, Victoria, 3052

Accounting policies

The following significant accounting policies have been adopted in the preparation and presentation of the financial report:

(a) Revenue recognition

Research Grants

When the Alliance receives government grants that are within the scope of AASB 1058 (being a transaction where the consideration paid to acquire an asset is significantly less than fair value principally to enable the Alliance to further its objectives), it performs an assessment to determine if the contract is 'enforceable' and contains 'sufficiently specific' performance obligations.

In cases where there is an 'enforceable' contract with a customer with 'sufficiently specific'

performance obligations, the transaction is accounted for under AASB 15 where income is recognised when (or as) the performance obligations are satisfied.

Member contributions

Income under AASB 1058 (being a transaction where the consideration paid to acquire an asset is significantly less than fair value principally to enable the Alliance to further its objectives). Member contributions are accrued on a quarterly basis as per Section 11 of the collaboration agreement and recognised as.

(b) Cash and cash equivalents

Cash comprises cash on hand and on demand deposits. Cash equivalents are short-term, highly liquid investments that are readily convertible to known amounts of cash, which are subject to an insignificant risk of changes in value and have a maturity of six months or less at the date of acquisition.

(c) Trade and Other Payables

Trade and other payables are initially measured at fair value on inception and then subsequently carried at amortised cost. They are recognised when the Alliance becomes obliged to make future payments resulting from the purchase of goods and services. The Alliance derecognises financial liabilities when, and only when, the Alliance's obligations are discharged, cancelled or have expired. The difference between the carrying amount of the financial liability derecognised and the consideration paid and payable is recognised in profit or loss.

(d) Goods and Services Tax (GST)

Revenue, expenses and assets are recognised net of the GST amount except:

- (i) where the amount of GST incurred is not recoverable from the taxation authority, it is recognised as part of the cost of acquisition of an asset or as part of an item of expense; or
- (ii) for receivables and payables which are recognised inclusive of GST.

The net amount of GST recoverable from, or payable to, the taxation authority is included as part of receivables or payables. Cash flows are included in the statement of cash flows on a gross basis.

(e) Critical accounting judgements and key sources of estimation uncertainty

In the application of the Alliance's accounting policies, which are described above, management may from time to time make judgements, estimates and assumptions about the carrying values of assets and liabilities that may not be readily apparent from other sources. The estimates and associated assumptions are based on historical experience and various other factors that are believed to be reasonable under key circumstances, the result of which form the basis of making the judgement. Key areas in which management has exercised judgement include the calculation of the carrying value of employee benefits.

(f) Impact of new and amended standards adopted

There were no new accounting standards or interpretations adopted in 2023 that had a significant effect on the Alliance.

2. Operating expenses	2023 \$'000	2022 \$'000
The following items of expense are included in the net surplus.		
(a) Remuneration of auditors		
Auditing the financial report	32	42
(b) Employee benefits expense		
Employee benefits expense	3,541	2,832
3. Trade and other receivables		
Accrued income	459	496
Trade and other receivables	3,111	1,549
	3,570	2,045
4. Trade and other payables		
Accrued expenses	766	1,115
Trade and other payables	590	1,363
	1,356	2,478
5. Unearned grant income		
Grants already committed and applicable to future periods	14,671	17,989
	14,671	17,989

6. Notes to statement of cash flows	2023 \$'000	2022 \$'000
(a) Reconciliation of cash		
For the purposes of the statement of cash flows, cash includes cash on hand and cash at bank, net of outstanding bank overdrafts.		
Cash at the end of the financial period as shown in the statement of cash flows is reconciled to the related items in the statement of financial position as follows:		
Cash	8,457	8,422
Term deposits	4,000	4,000
	12,457	12,422
(b) Reconciliation of net surplus to net cash flows from operating activities		
Net surplus	-	-
Changes in net assets and liabilities:		
Increase in assets:		
Trade and other receivables	(1,525)	(428)
(Decrease)/increase in liabilities:		
Trade and other payables	(1,122)	1,614
Other current liabilities (Grants)	(3,318)	8,157
Net cash from operating activities	(5,965)	9,343

7. Events after the reporting period

The directors are not aware of any other matter of circumstance which has arisen since the end of the financial year which has significantly affected or may significantly affect the operations of Melbourne Genomics Health Alliance, results of those operations or the state of affairs of Melbourne Genomics Health Alliance in subsequent financial years.

Directors' declaration

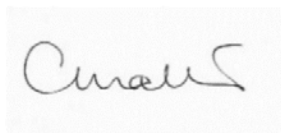
As detailed in note 1 to the financial statements, the Alliance is not a reporting entity because in the opinion of the Directors there are unlikely to exist users of the financial report who are unable to command the preparation of reports tailored so as to satisfy specifically all of their information needs. Accordingly, this special purpose financial report has been prepared to satisfy the Directors' reporting requirements.

The Directors declare that:

- (a) In the Directors' opinion, there are reasonable grounds to believe that the Alliance will be able to pay its debts as and when they become due and payable; and
- (b) In the Directors' opinion, the attached financial statements and notes thereto comply with accounting standards in Note 1 and give a true and fair view of the financial position and performance of the Alliance.

Signed in accordance with a resolution of the Directors.

On behalf of the Directors



Catherine Walter AM

Director (Board Chair)

20 June 2024



Adam Horsburgh

Finance, Audit and Risk Committee (Chair)

20 June 2024



Independent Auditor's Report to the Members of Melbourne Genomics Health Alliance

Opinion

We have audited the financial report, being a special purpose financial report of Melbourne Genomics Health Alliance ("MGHA" or the "Entity"), which comprises the statement of financial position as at 31 December 2023, the statement of profit or loss and other comprehensive income, statement of changes in equity and statement of cash flows for the year then ended, and notes to the financial statements, including a summary of material accounting policies, and the declaration by the Directors.

In our opinion, the accompanying financial report presents fairly, in all material respects, the Entity's financial position as at 31 December 2023, and of its financial performance and its cash flows for the year then ended in accordance with the Funding Agreement between the Department of Jobs, Precincts and Regions and MGHA.

Basis for Opinion

We conducted our audit in accordance with Australian Auditing Standards. Our responsibilities under those standards are further described in the *Auditor's Responsibilities for the Audit of the Financial Report* section of our report. We are independent of the Entity in accordance with the ethical requirements of the Accounting Professional and Ethical Standards Board's APES 110 *Code of Ethics for Professional Accountants (including independence standards)* (the Code) that are relevant to our audit of the financial report in Australia. We have also fulfilled our other ethical responsibilities in accordance with the Code. We believe that the audit evidence we have obtained is sufficient and appropriate to provide a basis for our opinion.

Emphasis of Matter – Basis of Accounting and Restriction on Distribution and Use

We draw attention to Note 1 to the financial report, which describes the basis of accounting which states that the financial report has been prepared in accordance with the recognition and measurement requirements by specified accounting standards for meeting the reporting requirements of the underlying funding agreement. The financial report has been prepared to assist the Entity to meet the financial reporting requirements of the Alliance under this agreement. As a result, the financial report may not be suitable for another purpose. Our report is intended solely for the Alliance and should not be distributed or used by parties other than the Alliance. Our opinion is not modified in respect of this matter.

The Directors of the Entity are responsible for the preparation of the financial report in accordance with Australian Accounting Standards to the extent described in Note 1 and for such internal control as the Directors determine is necessary to enable the preparation of the financial report that is free from material misstatement, whether due to fraud or error.

In preparing the financial report, the Directors are responsible for assessing the ability of the Entity to continue as a going concern, disclosing, as applicable, matters related to going concern and using the going concern basis of accounting unless management either intend to liquidate the Entity or to cease operations, or has no realistic alternative but to do so.

The Directors are responsible for overseeing the Entity's financial reporting process.

Auditor's Responsibilities for the Audit of the Financial Report

Our objectives are to obtain reasonable assurance about whether the financial report as a whole is free from material misstatement, whether due to fraud or error, and to issue an auditor's report that includes our opinion. Reasonable assurance is a high level of assurance, but is not a guarantee that an audit conducted in accordance with the Australian Auditing Standards will always detect a material misstatement when it exists. Misstatements can arise from fraud or error and are considered material if, individually or in the aggregate, they could reasonably be expected to influence the economic decisions of users taken on the basis of this financial report.

Liability limited by a scheme approved under Professional Standards Legislation.

Member of Deloitte Asia Pacific Limited and the Deloitte Organisation.

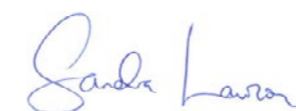
Responsibilities of the Directors for the Financial Report

As part of an audit in accordance with the Australian Auditing Standards, we exercise professional judgement and maintain professional skepticism throughout the audit. We also:

- Identify and assess the risks of material misstatement of the financial report, whether due to fraud or error, design and perform audit procedures responsive to those risks, and obtain audit evidence that is sufficient and appropriate to provide a basis for our opinion. The risk of not detecting a material misstatement resulting from fraud is higher than for one resulting from error, as fraud may involve collusion, forgery, intentional omissions, misrepresentations, or the override of internal control.
- Obtain an understanding of internal control relevant to the audit in order to design audit procedures that are appropriate in the circumstances, but not for the purpose of expressing an opinion on the effectiveness of the Entity's internal control.
- Evaluate the appropriateness of accounting policies used and the reasonableness of accounting estimates and related disclosures made by the Directors.
- Conclude on the appropriateness of the Directors' use of the non-going concern basis of accounting. Our conclusions are based on the audit evidence obtained up to the date of our auditor's report.
- Evaluate the overall presentation, structure and content of the financial report, including the disclosures, and whether the financial report represents the underlying transactions and events in a manner that achieves fair presentation.

We communicate with the Directors regarding, among other matters, the planned scope and timing of the audit and significant audit findings, including any significant deficiencies in internal control that we identify during our audit.


DELOITTE TOUCHE TOHMATSU


Sandra Lawson
Partner
Chartered Accountants
Melbourne, 20 June 2024

Melbourne Genomics Health Alliance

Alliance members

The Royal Melbourne Hospital
The Royal Children's Hospital
The University of Melbourne
WEHI
Murdoch Children's Research Institute
CSIRO
Australian Genome Research Facility
Peter MacCallum Cancer Centre
Austin Health
Monash Health

Melbourne Genomics Health Alliance

c/-WEHI,
1G Royal Parade, Parkville VIC 3052
melbournegenomics.org.au
enquiries@melbournegenomics.org.au

Melbourne Genomics acknowledges the Wurundjeri people of the Kulin Nation, on whose lands we work, and all First Nations peoples across Victoria. We pay respect to Elders past and present. We also acknowledge the First Nations health professionals, researchers and leaders who are shaping the future of genomic medicine.

