Melbourne Genomics
Health Alliance

# From possibility to practice

## Year in Review 2022

Alliance members

The Royal Melbourne Hospital
The Royal Children's Hospital Melbourne
The University of Melbourne
WEHI brighter together
murdoch children's research institute
CSIRO
agrf
Peter Mac Peter MacCallum Cancer Centre Victoria Australia
Austin Health
Monash Health

Supported by

Victoria State Government

Melbourne Genomics/Rodney Dekker

# Contents



**Genomic sequencing helped diagnose Yusuf. But it could also inform the care he gets over the entire course of his life.**

Our clinical platform GenoVic not only makes genomic testing easier; it stores genomic records so doctors can use them to inform future patient care.

***Six medical laboratories now use GenoVic to perform accredited tests.***

This year we made huge upgrades to its security and usability. Watch Yusuf's story at **diagnosisday.org.au**.



# About Melbourne Genomics

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

In 2013, leading Victorian hospitals, research and academic institutions joined forces to create the **Melbourne Genomics Health Alliance**.

Our studies have shown how genomics can provide answers, change treatment, and save lives. Now we're working to make genomic medicine a reality for all Victorians.

## The start of our story...

From 2013-2019, Melbourne Genomics sought to establish when genomics could enable better care for Victorian patients.

We conducted clinical studies across a range of health conditions, supported by the Victorian Government.

Overall, 19x more patients received an informative result from genomics than from usual care. Among cancer and rare disease patients, 42% received an informative result; 1 in 2 of those had a change in care because of it.

Meanwhile, we developed genomics education programs to build expertise within the health workforce. We also built a world-first clinical platform to support the end-to-end workflow of a genomic test and securely manage the big data it generates.

In 2021, we began a new program to embed genomics in Victoria's health system.

We laid the foundations for three streams of work:

- **Genomics in Practice** tests frameworks and models of care to guide health services in implementing genomics.
- **Genomics Workforce** builds the expertise of those who conduct genomic tests and those who use the tests in patient care.
- **Genomic Information Management** ensures genomic data is used effectively in healthcare, through the evolution and uptake of the GenoVic data platform.

Visit **melbournegenomics.org.au** for studies and reports

# Highlights from 2022

**The GenoVic clinical platform has been used to conduct more than**

## 15,000 genomic tests.

**718 professionals built their knowledge as a result of our genomics education.**

**A group of healthcare leaders and change experts is informing the implementation of genomics in hospitals.**

**Research with doctors, parents and geneticists identified ways to increase the use of genomic tests in paediatric care.**

**Three projects were selected to road-test solutions to the challenges of using genomics in healthcare.**

**A genomic snapshot of eight 'superbugs' was conducted across Victoria.**

**The Diagnosis Day mini-series followed six Victorian families with rare genetic conditions, to highlight the impact of genomic testing.**



# Introduction

Research opens our eyes to new possibilities. Turning that possibility into practice can be hard work.

Our last program showed how Victorians could benefit from genomics, through better diagnosis, targeted treatment, and preventive healthcare. In some ways, our work is now harder: finding the right ways to bring genomics into healthcare around the state.

We designed projects to tackle known barriers to implementing genomics: clinical frameworks and guidelines, knowledge and confidence, access and equity, and the safe and appropriate use of genomic data. But as we test these out, we find new and interconnected challenges.

For example, what are the most effective ways for regional hospitals to bring genomics into care? What is stopping doctors from using tests that are already Medicare funded? How do we bring genomic information into other health records? What is known about how First Nations and CALD families access genetics services?

Our final program seeks to answer these questions and more. We are road-testing different models of care and access to genomic expertise; co-designing solutions with clinicians and patients; making quality genomics education easy to access; and enabling genomic data to be used effectively, stored securely, and shared when needed to improve patients' health.

This work is grounded in implementation science: to test models, methods and strategies that support clinicians when they are ordering genomic tests. We are effectively creating a roadmap that makes it easier for Victorian health services to take on genomics.

I hope you enjoy this Year in Review.

**Prof Clara Gaff**
*Executive Director, Melbourne Genomics*

**Patrick's body can't process the protein phenylalanine, which is found in most foods.**

Testing after birth detected this condition before it led to brain damage. Now Patrick's on a restricted diet, but he lives a healthy, happy and very active life. Watch his story at **diagnosisday.org.au**.

***Genomic testing will diagnose a wider range of childhood conditions and give families access to life-changing treatments.***

We're working with paediatricians, geneticists and parents to expand the use of this test across Victoria.

Melbourne Genomics/Rodney Dekker

# Our program

## Streams

### Genomics in Practice

- Paediatrics Statewide
- Cancer Statewide
- Controlling Superbugs Statewide
- Clinical Change
  - Transplant
  - Nephrology
  - Dementia
- Equitable access
  - Understanding access
  - Understanding priorities
- Hospital implementation
- Immersive implementation

### Genomics Workforce

- Education for those using genomic tests
- Education for those conducting genomic tests
- Enduring education

### Genomic Information Management

- Innovation
- Data access and reuse
- Privacy and security
- Service delivery

## Projects

## Outcomes

Health services are ready and able to offer genomics.

Genomics is used more widely and equitably in clinical care.

Clinical, diagnostic & data science professionals are equipped to practise genomics.

Victoria has embedded pathways to build the genomic expertise of its workforce.

Our clinical genomics platform supports the safe provision of genomic health investigations.

Genomic data is used for clinical care and research in an ethical, trusted and secure way.

**When Josh would stop breathing, doctors put it down to behavioural issues. He was left to wait hours in emergency rooms.**

Genomic testing finally confirmed a diagnosis of Pitt-Hopkins Syndrome. This informed a management plan between his hospital's paediatrics, neurology, and respiratory team: ensuring when Josh needs help, he gets it fast.

***We're developing a framework to guide hospitals in implementing genomics safely and effectively.***

This can greatly reduce the time it takes to diagnose complex conditions like Pitt-Hopkins.

Watch Josh's story at **diagnosisday.org.au**.

Melbourne Genomics/Rodney Dekker



# Progress report

# Finding the right model for each health service

## Are hospitals ready for genomics yet?

Genomics requires multi-level change. Hospitals tell us they need guidance to navigate that change safely and effectively.

Informed by a group of healthcare change experts, we are building a framework and maturity model to guide hospital leaders in implementing genomics. Our framework addresses quality, safety, clinical effectiveness, workforce, and consumer participation.

We are also road-testing different models of care through our statewide and clinical change projects. This will help each health service choose a model that's right for them.

Melbourne Genomics/Zayne D'Crus

## Testing without travel: what's right for regional patients?

Genomics can make cancer treatment more precise, but regional Victorians commonly travel to larger tertiary hospitals to get a test and their results.

Our Cancer Statewide project aims to make it easier for oncologists to use genomics in cancer care. It compares three models of care across six regional and metropolitan hospitals. The first model will see oncologists continue to request genomic tests and receive reports from the Peter MacCallum Cancer Centre. The second and third models test the value of providing oncologists with additional support to interpret test results and use them in patient care: either through telehealth consultations with Peter Mac genomics specialists, or by training a local genomics 'superuser' based at the hospital.

This project will inform the broader rollout of genomics in oncology – meaning more Victorians can access precision treatment for cancers, closer to home.

**Eddy has had extensive surgeries, which leave him vulnerable to superbug infections when he goes to hospital.**

Hospitals can stop a superbug transmissions before they reach patients like Eddy – by sequencing a microbe's genome to understand where it came from, and overlaying that with patient movement data to understand how it's moving around the hospital.

It's detailed detective work, with the potential to transform the way hospitals maintain and extract information from their electronic medical records.

## Superbug super-sleuths resolve supersized superblock

Superbugs are antibiotic-resistant microbes, considered one of the world's biggest health threats.

In principle, a hospital can track superbug transmission by sequencing the bug's genome, and then cross-reference it with patient movement data. Infection control teams can determine what type of bug it is, who's carrying it, and how it's moving through the hospital, thus stopping a single transmission from becoming an outbreak.

In practice, we hit a few roadblocks.

The Controlling Superbugs project team was able to rapidly sequence and analyse superbug genomes – but extracting patient movement data from hospitals turned out to be even harder. Each hospital has its own way of storing patient movement data in its electronic medical record (EMR) system. This meant days wasted trying to reconcile differences in datasets.

The team has developed a standardised process that can extract patient movement data across multiple hospitals, so long as essential data hygiene steps are followed. This will enable hospitals to control superbug outbreaks in real time, protecting vulnerable patients.

The Controlling Superbugs project also systematically collected data from pathology labs to produce a genomic snapshot of the most significant antibiotic-resistant microbes in Victoria.



**A genomic test changed the course of Susan's cancer journey.**

**It enabled her oncologist, Dr Wasek Faisal, to prescribe a targeted immunotherapy course that eliminated her tumours.**

***Regional hospitals are exploring ways to access genomics expertise without requiring patients like Susan to travel for testing.***

**We are testing different models of embedding genomics in regional oncology services: including telehealth consultations and local 'superusers' with genomics expertise.**

Melbourne Genomics/Terry Hope

# Solutions for the real world

## Solving old problems while breaking new ground

This year saw the start of three projects that road-test practical solutions to the challenges of implementing genomics.

The first project offers genomic testing to people receiving **liver and kidney transplants**, to prevent adverse drug reactions and inform pre- and post-operative care. This project will test how proactive genomic sequencing can inform long-term patient care.

The second project explores how to bring genomics into the care of people with **early-onset dementia and cognitive disorders**. Genetic counsellors will join the neurology teams that support patients and families before and after a diagnosis, helping people to understand and act on information about their health and risk factors in a timely way.

The final project supports health services to provide genomic testing for **kidney diseases**, now covered by Medicare. Local 'kidney genomics champions' will be trained at both larger and peripheral hospitals, while a statewide multidisciplinary team of experts will be available to discuss challenging cases.

A consistent model of change will be applied to each project. Lessons from their implementation will guide other health services in introducing genomics across different disease areas.

**For years, Scott endured the pain of kidney stones. What he found harder to endure was not knowing why.**

Genomic testing pinpointed the cause of Scott's kidney disease, and also suggested a simple way to manage it – high doses of vitamin B6. Watch his story at **diagnosisday.org.au**.

***Evidence from Melbourne Genomics contributed to Medicare funding for genomic testing for kidney disease.***

The next step is to ensure health services can keep up with demand, by trialling ways to access genomic expertise.

Melbourne Genomics/Rodney Dekker



## The gap between knowing and doing

While there's a growing interest in genomic testing within many medical specialties, doctors say they don't always have the confidence to use it in patient care.

### Building knowledge

This year, 718 professionals built their knowledge through our genomics education. This included:

- making genomics education readily accessible, through self-directed and blended learning courses
- taking genomics to regional clinicians, through interactive in-person workshops
- new speciality workshops and webinars: using genomic testing for suspected familial cancer and for paediatric oncology; and assisting paediatricians in interpreting exome reports and talking with parents about genomic testing
- advanced training in variant interpretation
- introducing genomics as a career option for data scientists, medical scientists and other professionals, through targeted webinars

Post-event surveys showed that 97% of doctors felt confident in at least one relevant genomic skill after clinical education.

### Putting knowledge into practice

We're also exploring ways for health professionals to put their knowledge into practice, with support available when it's needed.

- Hospitals identified 'immersive implementation' projects that can build genomics expertise through supervised internships or by tackling barriers to implementation.
- Other projects are trialling alternative ways for doctors to seek advice on using genomics. This includes upskilling local 'genomics champions' who can assist their colleagues; placing genetic counsellors within specialist clinics; using telehealth consultations to explain genomic testing and deliver results to patients; and building connections between geneticists and regional medical specialists, to open the door to future discussion of cases.

**718**

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

**97%**

***of doctors felt confident in at least one genomic skill after clinical education.***

# One platform, 15,000+ tests

Managing the big data from genomic tests is one of the most critical challenges facing the digital health system. Our clinical platform – currently called GenoVic – was created to tackle this challenge head on.

GenoVic has supported over 15,000 clinically-accredited genomic tests, with a third of those in 2022. Six medical laboratories now use the platform to test for both inherited conditions and cancer.

Major upgrades in 2022 made GenoVic a platform that can be used by health services in multiple jurisdictions across Australia.

## GenoVic now supports data sharing between laboratories.

With consent from patients, laboratories can now share their clinical genomic data with other labs (to ensure continuity of care for patients) or with researchers (to power new insights and advances). This critical feature is supported by strong data governance and privacy procedures, developed through extensive stakeholder consultations.

## Onboarding new users is easier.

Austin Health became the sixth pathology provider to use GenoVic, while Alfred Health added a new workflow in the system during the year. Both laboratories chose GenoVic for its process efficiencies, leading-edge genomic tools, and data sharing capacity.

## Software improvements were delivered more predictably and efficiently.

Twelve new GenoVic releases were delivered in the past year.

## A new portal gives users more control

Laboratories can now audit their actions in the platform, and more easily manage individual users and their level of access.

## Cybersecurity ramped up.

GenoVic was audited and tested by independent experts to ensure it complies with best-practice security measures. Security is an ongoing journey, and each audit helps us take steps to stay ahead of cyberthreats.

## A health integration engine was introduced.

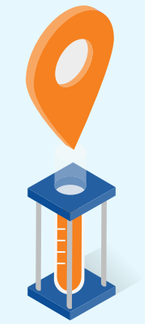
This enables GenoVic to communicate with electronic medical records such as EPIC: meaning it will be easier for doctors to use their existing records system to request genomic tests and receive reports.



*GenoVic has supported over*

**15,000**

*genomic tests, with a third of those in 2022.*



*For every*

**1,000**

*tests conducted through GenoVic, a lab can save the equivalent of two full-time employees, freeing up their time for testing rather than administration.*

*GenoVic now enables a lab to share a patient's genomic data with another lab, to conduct a new test if the patient needs one.*

# With patients at the centre

## There's evidence and funding. What's missing?

The evidence is there: genomic tests can provide answers about many different childhood conditions. The funding is there: Medicare covers genomic testing for children with specific characteristics. What's missing is the widespread use of these tests.

In 2022, we conducted research with paediatricians, parents and geneticists to understand what barriers are limiting the use of genomic tests. Three key issues emerged. First, doctors don't always know when genomic sequencing is the right test to use. Second, it can be difficult for paediatricians to access geneticists for advice, especially if it's needed during a patient consultation. Third, genomic sequencing is a complex test to explain to parents.

Doctors, parents, geneticists and rare disease support groups will now engage in a co-design process to create and test solutions. These include ways to enable timely access to genetic experts, education and on-the-job support for paediatricians, and resources and referrals for patients and parents.

## What story do the numbers tell?

Many people face barriers to healthcare relating to language, culture and discrimination. Genomics is still new: we can address these barriers at every step of its implementation.

This begins by understanding who's currently accessing genetics services, who isn't, and why.

By auditing records from Victorian genetic services, we may be able to see whether First Nations and culturally and linguistically diverse (CALD) families are accessing these services at an expected and equitable rate. Data will be analysed for common patterns and themes. This work will be led by Dr Angeline Ferdinand at the University of Melbourne.

Meanwhile, we are supporting the establishment of a Victorian node for the MRFF-funded National Indigenous Genomics Network, led by Prof Alex Brown. Its work will include engaging with Victorian Aboriginal community-controlled health organisations, to better understand their genomic health priorities and capacity requirements, as well as barriers to accessing genetics services and models that can improve access.



## "Language when you're receiving a diagnosis is so important," says Deanna.

"'Go home and make memories' is a phrase they used. It told me there's nothing you can do as a parent."

Deanna and Andrew's son was diagnosed with Niemann-Pick Type C Syndrome. They shared their experience at **diagnosisday.org.au** to help clinicians understand the challenges of communicating a potentially fatal diagnosis.

***Real-life stories and clinical case studies underpin our education events for paediatricians, oncologists and other medical specialists.***

The Diagnosis Day stories informed a new workshop for paediatricians on how to discuss genomic testing with families.

Melbourne Genomics/Rodney Dekker

## Six families search for answers

A study provides the evidence for change. A model shows people how to make the change. But a story makes people want to change.

That's why we teamed up with the Genetic Support Network of Victoria to tell the story of six families living with rare genetic conditions. Together we created a seven-episode series, Diagnosis Day, that follows the families on their search for answers, the moment of diagnosis, and the impact of diagnosis on their future.

These stories are a powerful advocacy tool. They help clinicians understand why diagnosis matters, and how to support families with rare and undiagnosed conditions.

Our Community Advisory Group, consumer advocates and genetic counsellors guided the entire process: from recruiting participants to developing interview questions, and reviewing scripts and rough cuts.

Diagnosis Day was released on LinkedIn and YouTube in November 2022, attracting well over 40,000 views. It is being shared widely within the rare disease sector, and has been used to support education for paediatricians who need to discuss genomic testing with families.

**Watch the series at diagnosisday.org.au.**

# Thinking nationally

Melbourne Genomics provides evidence to inform national decisions about genomics. Our members also support and enhance nationwide genomics projects.

**$1.9M in value was generated for Victoria in 2022 through Medicare rebates for genomic testing of Victorians.**

Melbourne Genomics studies informed the decision to fund testing for childhood syndromes kidney disease and heritable cardiomyopathies.

**We are supporting the Victorian node of a National Indigenous Genomics Network.**

Led by First Nations researchers, the network will help ensure genomics explicitly benefits Aboriginal and Torres Strait Islander peoples.

**More than $16M was awarded to Melbourne Genomics member organisations through the Genomics Health Futures Mission.**

This is part of the Federal Government's Medical Research Future Fund. Victorian researchers including our executive director Clara Gaff are also supporting two national projects: ethical governance for genomic data, and a National Indigenous Genomics Network.

**We brought a clinical perspective to national policy discussions on the management of genomic data.**

These discussions were led by Australian Genomics to create a federated ecosystem of genomic, health and research solutions, linked by interoperability and guided by shared standards. Our clinical data platform, GenoVic, will be a vital component of the nationwide ecosystem.



# The people taking genomics forward

# Our Board of Directors

The Melbourne Genomics Board comprises senior leaders from each of the 10 Alliance members, plus an Independent Director and an Independent Chair.

The Board includes a Finance, Audit and Risk Committee, and a Business Development and Commercialisation Committee.

The Board met four times in 2022, providing invaluable guidance on strategic planning, allocation of resources, and commercialisation of program assets.

**Catherine Walter AM**
Independent Chair
Appointed: Jul 2016

**Prof Christine Kilpatrick**
Chief Executive,
The Royal Melbourne Hospital
Appointed: Oct 2015

**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

**Prof Kathryn North AC**
Director, Murdoch Children's Research Institute
Appointed: Oct 2015

**Dr Rob Grenfell**
Health Director of the Health and Biosecurity Business Unit, CSIRO
Appointed: Dec 2016
Retired from Board: Sep 2022

**Lynne Cobiac**
Health Director of the Health and Biosecurity Business Unit, CSIRO
Appointed: Sep 2022

**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

**Adam Horsburgh**
Chief Executive Officer,
Austin Health
Appointed: Mar 2017

**Andrew Stripp**
Chief Executive, Monash Health
Appointed: May 2016

**Dr Dan Grant**
Independent Director
Appointed: Dec 2021

**Visit our website for profiles of Board Members**

## Finance, Audit and Risk Committee

**Adam Horsburgh (Chair)**
Chief Executive Officer,
Austin Health
Appointed: May 2021

**Lucy Franzmann**
Chief Finance Officer, Peter MacCallum Cancer Centre
Appointed: May 2021

**Joh Kirby**
Head – Governance,
Risk and Compliance, WEHI
Appointed: May 2021

**Prof Matt Sabin**
Chief Medical Officer and Executive Director – Medical Services and Clinical Governance
The Royal Children's Hospital
Retired from committee: Aug 2022

## Business Development and Commercialisation Committee

**Dr Dan Grant**
Chair
Appointed: Dec 2021

**Dr Angus Henderson**
Director, Enterprise Strategy & Development, Monash Health
Appointed: May 2022

**Dr Anne-Laure Puaux**
Head, Biotechnology and Commercialisation, WEHI
Appointed: May 2022

**Dr Janet Fox**
Business Development Manager, CSIRO
Appointed: May 2022

**Joe Baini**
Chief Executive Officer, AGRF
Appointed: May 2022

**Kathryn Thomas**
General Manager,
Victorian Clinical Genetics Services
Appointed: May 2022
Retired from Committee: Aug 2022



# Community Advisory Group (CAG)

This group advises on how we involve consumers and communities in our program.

Its members are champions of consumer rights in healthcare.

The group met four times in 2022: advising when community engagement was needed for a project, how to approach it, and whose voices must be heard.

**Jane Bell (Chair)**
Appointed: Nov 2016
Retired from CAG: May 2022

**Kellie-Ann Jolly (Chair)**
Appointed: Aug 2022

**Louisa Di Pietro**
Appointed: Jan 2014

**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

**Pamela Williams**
Appointed: May 2018
Retired from CAG: May 2022

**Visit our website for member profiles**

# Hospital Implementation Reference Group

This group was convened in 2022 to provide strategic advice on the implementation and sustained use of genomics in Victorian hospitals.

**Dr Jason Goh**
Director Medical Services,
Monash Health
Appointed: Mar 2022

**A/Prof Tom Connell**
Chief of Medicine,
Royal Children's Hospital
Appointed: Mar 2022

**Dr Charlotte McArthur**
Executive Director – Strategy, Performance and Improvement,
Austin Health
Appointed: Mar 2022
Retired from group: Apr 2022

**Rachel Meehan**
Executive Director – Strategy, Performance and Improvement,
Austin Health
Appointed: Apr 2022

**Dr David Speakman**
Chief Medical Officer, Peter MacCallum Cancer Centre
Appointed: Mar 2022

**Prof Kirsty Buising**
Medical Director of Medical Services, Royal Melbourne Hospital
Appointed: Mar 2022

# Experts across and beyond Victoria

Every Melbourne Genomics project is a collaboration with leading clinicians, researchers and academics from our Alliance members and further afield.

Here are some of the external experts who worked alongside our team in 2022.

## Genomics in Practice stream

The **Paediatrics Statewide** project has benefited from the expertise of **Dr Natasha Brown** of the Murdoch Children's Research Institute, **A/Prof Michael Fahey** from Monash Health, **Heather Renton** of SWAN and paediatrics trainee **Calder Hammill**. Experts from the University of Melbourne's MISCH Hub – **Jennifer Bibb**, **Victoria Palmer**, **Roxanne Kritharidis**, **Erin Davis**, **Cathy Vaughan** and **Julie Simpson** – informed the co-design processes used in this project.

The **Cancer Statewide** project is led by **Prof Jayesh Desai** and **Dr Kortnye Smith** of Peter MacCallum Cancer Centre, together with **Dr Laura Forrest**, **Prof Stephen Fox** and **Dr Lavinia Tan** of Peter Mac, **A/Prof Natalie Taylor** from UNSW, and **Dr Michelle Tew** from the University of Melbourne.

Infectious disease experts from Austin Health lead the **Controlling Superbugs** project: **Prof Lindsay Grayson**, **Prof Rinaldo Bellomo**, **A/Prof Jason Kwong**, **Dr Norelle Sherry** and **A/Prof Natasha Holmes**. Additional expertise comes from **Dorothy Ling**, **Sean Mace**, **Jordana Flude** and **Kartik Kishore** at Austin Health.

Three new Clinical Change projects were conceived by Alliance clinicians. The **Nephrology** project is led by **Dr Kushani Jayasinghe** of Monash Health, the **Dementia** project by **Dr Aamira Huq** at the Royal Melbourne Hospital, and the **Transplant** project by **Prof Paul James** at the Royal Melbourne Hospital. Each project is supported by experts from across the Alliance.

**A/Prof Cate Kelly** is advising on governance and guiding our **Hospital Implementation** project, bringing together a wide range of healthcare leaders, implementation scientists and other experts.

**Prof Angeline Ferdinand** from the University of Melbourne will lead research into how Aboriginal, Torres Strait Islander and CALD patients access genetic services in Victoria. **Prof Alex Brown** and **Louise Lyons** from the National Indigenous Genomics Program lead engagement with Aboriginal community-controlled health organisations.



## Genomics Workforce

Our genomics education programs were informed by an advisory group that includes **Prof John Christodoulou** from The University of Melbourne, **A/Prof Amy Gray** from The Melbourne Academic Centre for Health, **A/Prof Femke Buisman-Pijlman**, **Dr Jennifer Weller-Newton** and **Prof Steve Trumble** from The University of Melbourne, and **Prof Stephen Fox** from Peter MacCallum Cancer Centre. **Michelle Barrett** from the VCCC Alliance, **A/Prof Catherine Quinlan** from the Royal Children's Hospital, **A/Prof Sebastian Lunke** from Victorian Clinical Genetic Service and **Dr Angus Henderson** from Monash Health also advised on an enduring education strategy.

**Dr Bryony Thompson** from The Royal Melbourne Hospital, **A/Prof Cathy Quinlan** and **Dr Dong Anh Khuong Quang** from The Royal Children's Hospital, **A/Prof Yoland Anthill** from Cabrini Family Cancer Clinic, Monash Health Familial Cancer Centre and Parkville Family Cancer Centre, **Dr Simon Bodek** from Austin Health, **Dr Victoria Bashay**, **Dr Kortnye Smith**, **Dr Lavinia Tan** and **Dr Huiling Xu** from Peter MacCallum Cancer Centre, **Dr Joep Vissers** from The University of Melbourne, and **Professor Zornitza Stark**, **Professor Tiong Tan**, **Professor Sue White**, **A/Prof Sebastian Lunke**, **Dr Naomi Baker**, **Dr Belinda Chong**, **Dr Dean Phelan**, **Lisette Curnow**, **Justine Elliot**, **Lyndon Gallacher** and **Elly Lynch** from Victorian Clinical Genetics Services made major contributions to our education events over the year.

The following people also presented or facilitated group work at events: **Prof Paul James** and **Dr Mark Cleghorn** from The Royal Melbourne Hospital, **Heather Chalinor** from Austin Health, **Dr Georgina Ryland** from Peter MacCallum Cancer Centre, **Dr Kushani Jayasinghe** from Monash Health, and Professor **David Amor**, **Professor Martin Delatycki**, **Dr Megan Ball**, **Dr Oliver Heath**, **Dr Chloe Stutterd**, **Dr Sok Kun Tae**, **Manny Jacobs**, **Nitzan Lang** and **Yana Smagarinksy** from Victorian Clinical Genetics Services.

## Genomic Information Management

GenoVic's ongoing evolution has been shaped by its key users across the Alliance and beyond. They include **Jane Lin** and **Adam Ivey** from Alfred Health; **A/Prof Kerryn Ireland-Jenkins**, **Dr Jonathon Clark** and **Dr Rishu Agarwal** from Austin Health; **Dr Giles Kelsey** and **Dr Bryony Thompson** from The Royal Melbourne Hospital; **George Cozaris** from The Royal Melbourne and Royal Women's Hospitals; and **Paul Kalitsis**, **Dr Simon Sadedin** and **Anthony Marty** from Victorian Clinical Genetics Services.

An Innovation and Prioritisation Committee helps ensure GenoVic can swiftly incorporate technological innovations. Its members include **Dr Angus Henderson** from Monash Health, **Dr David Hansen** from CSIRO, **Revital Rosenburg** from Murdoch Children's Research Institute, **Dr Vineesh Khanna** from the Victorian Department of Health, **Dr Cath Moore** from the Australian Genome Research Facility, and **Prof Paul James** from The Royal Melbourne Hospital.

## Program team

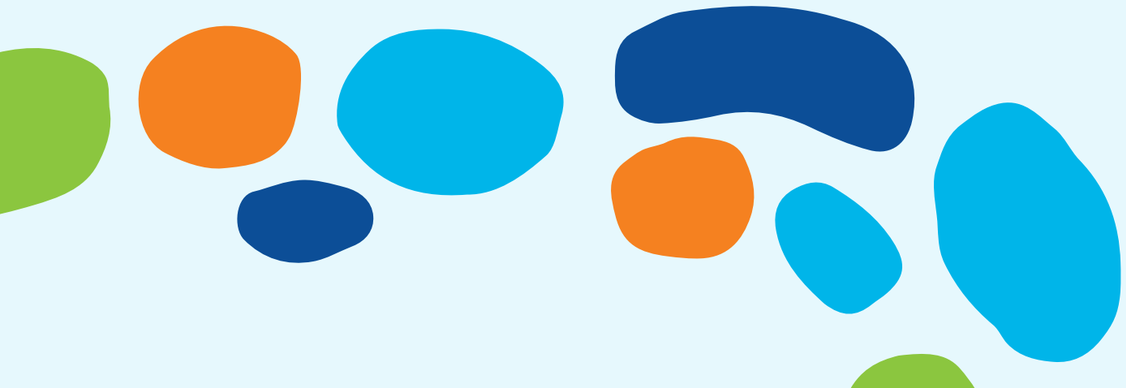
Our support team continues to evolve to meet the needs of the program.

The **Genomic Information Management** team worked intelligently and cohesively to strengthen GenoVic as a product, improve user experience and support, and engage a broad range of stakeholders. The team added health system integration and cloud software engineering to its already-impressive skillset. Six team members became certified health informaticians, greatly increasing their ability to help laboratories onboard and customise GenoVic.

The **Genomics in Practice** team grew considerably. Team members with expertise in clinical practice, research and project management are supporting implementation and evaluation of the complex projects led by Alliance member organisations. A key focus this year was supporting ethics and governance submissions for all projects.

The **Genomics Workforce** team further developed its capability to deliver a range of online, hybrid and in-person education activities. The team grew to include more learning design and event management expertise.

A business development manager joined us this year, working across all streams to ensure the sustainability of our clinical genomics system and education assets. Meanwhile, ongoing investment in evaluation and communication helps us capture the right information and use it to tell a compelling story.





# Recognising brilliance

**Dr Natalie Thorne** of Melbourne Genomics was recognised as one of 25 Brilliant Women in Digital Health in 2022.

The Brilliant Women awards, presented by Telstra Health, recognise women who have made outstanding achievements in digital health and aged care.

Natalie was celebrated for more than a decade's contribution to IT systems that enable genomic testing in healthcare, most significantly the GenoVic clinical system that's now used by six leading laboratories.

**Dr Kushani Jayasinghe**, Flagship clinician on the Genetic Kidney Disease Clinical Project, received the American College of Medical Genetics and Genomics (ACMG) Foundation's 2022 Richard King Award for Best Publication by a Trainee.

The award highlights articles published by trainees who were either a first or corresponding author in the ACMG's official journal, Genetics in Medicine.

Dr Jayasinghe's paper on the work of the Kidney Flagship – **Clinical impact of genomic testing in patients with suspected monogenic kidney disease** – was considered to have the most merit out of all the articles published by trainees.

A paper from our **Superbugs Flagship** was recognised at IDWeek, the annual meeting of several leading infectious disease groups.

The 2021 paper – **Pilot study of a combined genomic and epidemiologic surveillance program for hospital-acquired multidrug-resistant pathogens across multiple hospital networks in Australia** – won the William Jarvis Award for the best international manuscript in the Infection Control and Hospital Epidemiology journal.

**Dr Amy Nisselle** was recognised in Trinity College's 2022 Oakleaf Awards for her contribution to the advancement of medical science through education.


Dr Natalie Thorne at the Brilliant Women in Digital Health Awards.

# Where you saw us in 2022

| Date | Details | Presenter |
|---|---|---|
| 22/04/2022 | Ethics of Paediatric Genomics Symposium, Genetic health professionals' and parents' perspectives on offering additional findings to children with hearing loss | Danya Vears |
| 22/04/2022 | Ethics of Paediatric Genomics Symposium, Offering genomic sequencing and additional findings to children with hearing loss | Lilian Downie |
| 22/04/2022 | Ethics of Paediatric Genomics Symposium, Offering additional findings in the Acute Care Genomics Program | Clara Gaff |
| 12/05/2022 | The Royal Australasian College of Physicians Congress 2022: A Climate for Change, Genomics: How it can transform your practice (panel) | Cathy Quinlan |
| 1/06/2022 | 2022 Digital Health Festival, Data challenges of genomic medicine | Natalie Thorne |
| 12/06/2022 | European Society of Human Genetics Annual Scientific Meeting 2022, Invited workshop: What are patients' experiences with being offered Opportunistic Screening? | Melissa Martyn |
| 12/06/2022 | European Society of Human Genetics Annual Scientific Meeting 2022, Invited oral: How do we deliver genomics education for all? | Clara Gaff |
| 21/06/2022 | Illumina Genomics Summit 2022, Using genomics to control the spread of pathogens | Benjamin Howden |
| 31/08/2022 | Australian Institute of Medical and Clinical Scientists' VIC Branch Annual General Meeting 2022, Medical scientist career discovery session (panel) | Amy Nisselle |
| 23/09/2022 | Human Genetics Society of Australasia's SA Branch 2022 Symposium, Invited presentation: Responding to Workforce Challenges – Insights from Research and Education | Clara Gaff |
| 24/11/2022 | Human Genetics Society of Australasia 45th Annual Scientific Meeting, Genomic data in the digital health system: how can it happen? (workshop) | Natalie Thorne |



# Peer-reviewed publications

"**Shariant platform: Enabling evidence sharing across Australian clinical genetic-testing laboratories to support variant interpretation**". Emma Tudini, James Andres, David M. Lawrence, Sarah L. King-Smith, Naomi Baker, Leanne Baxter, Jogn Beilby, Bruce Bennetts, Victoria Beshay, Michale Black, Tiffany F. Boughtwood, Kristian Brion, Pak Leng Cheong, Michael Christie, Jogn Christodoulou, Belinda Chong, Kathy Cox, Mark R. Davis, Lucas Dejong, Marcel E. Dinger, Kenneth D. Doig, Evelyn Douglas, Andrew Dubowsky, Melissa Ellul, Andre Fellowes, Katrina Fisk, Cristina Fortuno, Kathryn Friend, Renee L. Gallagher, Song Gao, Emma Hacket, Johanna Hadler, Michael Hipwell, Gladys Ho, Georgina Hollway, Amanda J. Hooper, Karin S. Kassahn, Rahul Krishnaraj, Chiyan Lau, Huong Le, Huei San Leong, Ben Lundie, Sebastian Lunke, Anthony Marty, Mary McPhillips, Lan T. Nguyen, Katie Nones, Kristen Palmer, John V. Pearson, Michael C.J. Quinn, Lesley H. Rawlings, Simon Sadedin, Louisa Sanchez, Andreas W. Schreiber, Emanouil Sigalas, Aygul Simsek, Julien Soubrier, Zornizta Stark, Bryony A. Thompson, James U, Cassandra G. Vakulin, Amada V. Wells, Cheryl A. Wise, Rick Woods, Andrew Ziolkowski, Marie-Jo Brion, Hamish S. Scott, Natalie P. Thorne, Amanda B. Spurdle, on behalf of the Shariant Consortium. American Journal of Human Genetics (2022) **https://doi.org/10.1016/j.ajhg.2022.10.006**

"**Clinicians' views and experiences with offering and returning results from exome sequencing to parents of infants with hearing loss**", Lauren Notini, Clara L. Gaff, Julian Savulescu and Danya F. Vears, Journal of Clinical Medicine (2022) **https://doi.org/10.3390/jcm11010035**

"**Multi-site implementation of whole genome sequencing for hospital infection control: A prospective genomic epidemiological analysis**", Norelle L. Sherry, Claire L. Gorrie, Jason C. Kwong, Charlie Higgs, Rhonda L. Stuart, Caroline Marshall, Susan A. Ballard, Michelle Sait, Tony M. Korman, Monica L. Slavin, Robyn S. Lee, Maryza Graham, Marcel Leroi, Leon J. Worth, Hiu Tat Chan, Torsten Seemann, M. Lindsay Grayson, and Benjamin P. Howden. The Lancet Regional Health – Western Pacific (2022). **doi: 10.1016/j.lanwpc.2022.100446**

"**Structured approaches to implementation of clinical genomics: A scoping review**", Helen L. Brown, Isabella A. Sherburn, Clara Gaff, Natalie Taylor and Stephanie Best, *Genetics in Medicine* (2022) **https://doi.org/10.1016/j.gim.2022.03.017**

## Other publications

"**Transforming the genomics workforce to sustain high value care**", Janet C. Long, Clara Gaff and Chrissy Clay, *Deeble Perspectives Brief* (2022) **https://ahha.asn.au/sites/default/files/docs/policy-issue/perspectives_brief_no_20_genomics_workforce_0.pdf**

Melissa Martyn, Amy Nisselle, Elly Lynch and Clara L. Gaff (2022), 'Chapter 1: Theories and models for genomics education and training' in Dhavendra Kumar **Genomic Medicine Skills and Competencies**, Academic Press.

Melbourne Genomics
Health Alliance

# Annual Report

A complete annual report, including an operations report and financial statements for the **year ended 31 December 2022**, will be available after the audit in June 2023.