Melbourne Genomics
Health Alliance

# Genomics in Victoria:
# What's changed in 10 years?

2013-2023

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
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Peter Mac Peter MacCallum Cancer Centre Victoria Australia
Austin Health
Monash Health

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# Genomic medicine maps the big data in our cells, to predict or diagnose illness and find the right way to treat it.

A genomic test begins by sequencing a patient's genome, which comprises over 23,000 genes. The genome is analysed by computer programs to identify any gene changes (called variants). Experts examine these variants – considering family history, medical history and existing evidence – to see if they can predict or explain specific health conditions.

## Contents




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Cover photography by Rodney Dekker

# Foreword

## The Hon. Mary-Anne Thomas MP

Minister for Health

**It is remarkable what Victoria has accomplished in genomic medicine over the past decade.**

Healthcare systems are complex, and it can be hard to drive change. We know it can take many years for medical discoveries and advances to reach clinical practice.

But just 10 years since its establishment, the Melbourne Genomics Health Alliance has succeeded in embedding advanced genomics techniques across Victoria's healthcare system.

Patients are receiving better treatment in many areas. Critically ill babies are receiving results from genomic tests in less than three days. And, most importantly, more and more Victorians can access cutting edge genomic medicine, through funding from Medicare and the Victorian Government.

These advancements mean we can tailor our healthcare to individuals – delivering better treatment, more effective care, and life saving interventions.

Formed in 2013, the Alliance brings together Victorian hospitals and institutions with a shared vision of using genomic medicine to benefit Victorian patients.

The Victorian Government has invested in the Alliance since then to support this vision. This includes $35 million for its final program working to equip Victoria with the tools, resources and evidence to support the widespread, safe and equitable adoption of genomics.

Because of the Alliance's great work, we've been able to identify when genomic medicine can offer the most benefit for Victorian patients. We have built a Victorian data platform that improves the efficiency of genomic testing and makes it scalable. And we are researching how health services can safely and effectively implement genomic models of care.

All of this was possible thanks to the hard work of the Alliance and its members.

**I congratulate and thank all members of the Alliance, their staff, and everyone across the sector who has worked hard to bring genomic medicine to all Victorians.**

# Bringing genomics to Victoria

## Prof. Clara Gaff

Executive Director, Melbourne Genomics

**It took 13 years to sequence the first human genome. Ten years after it was released, leaders from Victoria's hospitals, research and academic institutions committed to making genomic medicine possible for Victorian patients.**

It became clear that Victoria needed trailblazers as well as discoverers. The benefits of genomics were known, but untested in the real world of healthcare.

It was equally clear that collaboration was the only path to success. Everyone's expertise was needed: from the doctors who would use a genomic test, to the scientists who could conduct the test, the informaticians and software developers who would build the technology behind it, the patients whose care would depend on it, and the researchers who could capture what everyone was learning.

So the Melbourne Genomics Health Alliance was formed in 2013. Its members set out a collaborative vision to bring genomics into healthcare, and each member made an equal financial contribution to this vision. The Victorian Government contributed funding in the following year; a joint investment that has continued ever since.

Ten years on, this report chronicles how Victoria introduced new technology and discovery into a complex health system. Given that it can take 17 years for research discovery to become clinical practice, change has happened remarkably fast.

I want to express my sincere gratitude to the leaders of our member organisations; the patients, clinicians, scientists and developers who participated in our work; and the Victorian Government ministers and advisors whose leadership made it possible.

The Alliance's program comes to an end in 2025. While incredible progress has been made, the work is not done yet, and the road ahead is exciting.

# What's changed in 10 years?

**Melbourne Genomics is an alliance of leading hospitals, research and academic institutions, working to bring genomics into routine healthcare.** Here is how genomic medicine has advanced in Victoria in the 10 years since the Alliance was formed.

**1** Genomics gave thousands of Victorians answers when other tests could not.

**2** Ten years ago, there were no Medicare-funded genomic tests. Now there are eight.

**3** Critically ill babies can get a rapid genomic test to inform their care.

**4** Genomics emerged as a powerful weapon in the fight against superbugs.

**5** Victoria worked out how to safely store and share genomic health data.

**6** Genomics is entering mainstream medicine.

**7** Over 3,000 health professionals in Victoria have some genomics expertise.

**8** Alliance members are at the forefront of genomic innovation.

**9** More people with genetic conditions are advocating for genomic medicine.

**10** There's an easier path between medical discovery and routine healthcare.

# 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 become 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.

Photo: Rodney Dekker

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**

# Genomics gave thousands of Victorians answers when other tests could not.

## Victoria led the world in providing real-time evidence for the use of genomic testing in patient care.

Genomic testing was just emerging in medicine in 2013. Studies at the time were retrospective: offering genomic tests to patients who had already been extensively investigated for years.

Melbourne Genomics was the first to compare genomic testing with other health investigations in real time. The Alliance needed to know whether genomics would lead to a better outcome than other kinds of tests, and for which health conditions.

Nearly 4,000 Victorians received genomic testing from 2014-2019 for conditions ranging from rare childhood syndromes to acute myeloid leukaemia, focal epilepsy, solid cancers, congenital deafness, neurological and immunological conditions, kidney disease and more.[1]

The results were astonishing. Across the board, 19 times more patients received an informative result from genomic testing than usual care. More than two in five cancer and rare disease patients got an answer about their condition; and doctors were able to change the care of over half of those patients as a result.

Some patients got simple but life-changing treatments. Others went from being terminal to cancer-free. Others were able to plan families with confidence, or knew how to provide their children with the right support to thrive. Many were spared painful and invasive tests or unnecessary medications.

Many of these studies were critical to the broader rollout of genomic medicine in Australia. They also showed how this complex test could be provided more widely, quickly and cost-effectively.

**19x more patients** received an informative result from genomic testing than from usual care.

**42% of cancer and rare disease patients** received an informative result. **1 in 2** of those patients had a change in care because of it.

**5x more patients with immunological conditions** received a diagnosis.

**2 in 5 patients with bone marrow failure** received a more accurate diagnosis.

1 Published results are available at **melbournegenomics.org.au**.


# Ten years ago, there were no Medicare-funded genomic tests. Now there are eight.

## Evidence from Victoria has been critical in getting several genomic tests funded by Medicare.

Medicare can only fund medical services whose safety, clinical effectiveness and cost effectiveness have been assessed by the Medical Services Advisory Committee. Rigorous evidence underpins each assessment.

Medicare began to fund genomic tests in May 2020. The first funded test was for **'childhood syndromes'** – conditions affecting children aged 10 and under, likely to be caused by a single gene change. Much of the evidence for this test came from a Melbourne Genomics project.[2] Crucially, this project also showed the value of re-analysing genomic data as new knowledge becomes available.

Seven more genomic tests are now Medicare-funded. Melbourne Genomics evidence was cited in applications for **kidney disease**[3] and **cardiomyopathies**[4] (our studies showed genomic testing was an effective diagnostic tool).

Starting in November 2023, Medicare will fund new genomic tests for blood cancers and childhood hearing impairment. Evidence from Melbourne Genomics was crucial in obtaining funding for **childhood hearing loss**: our study showed that genomic testing tripled the diagnosis rate and changed the care of 90% of children who were diagnosed.[5]

Early genomic sequencing for children with suspected monogenic conditions resulted in **five times more diagnoses**, for **less than half the cost** of usual testing and care.

**One in two patients** with genetic kidney disease got an informative result, and **one in four** got more precise care.

Finding the cause of **severe hearing loss in babies** tripled with genomic testing.

2 "A prospective evaluation of whole-exome sequencing as a first-tier molecular test in infants with suspected monogenic disorders", Stark, Z.,et.al, *Genetics in Medicine* (2016) doi:10.1038/gim.2016.1

3 "Clinical impact of genomic testing in patients with suspected monogenic kidney disease", Jayasinghe et.al, Genetics in Medicine (2020) **https://doi.org/10.1038/s41436-020-00963-4**

4 "Prospective evaluation of the utility of whole exome sequencing in dilated cardiomyopathy", Ramchand et.al., Journal of the American Heart Association (2020) doi.org/10.1161/JAHA.119.013346

5 "Exome sequencing for isolated congenital hearing loss: a cost-effective analysis", Downie et.al., The Laryngoscope (2020) **https://doi.org/10.1002/lary.29356**

# 3

# Critically-ill babies can get a rapid genomic test to inform their care.

**Victoria pioneered rapid genomic sequencing for critically-ill babies, showing it could deliver results up to ten times faster.**

In 2015, it took around 136 days to receive a genomic test result. Children in intensive care need answers much faster than that.

Melbourne Genomics members formed a 'rapids team', bringing together specimen reception staff, diagnostic laboratory scientists, bioinformaticians, variant curators, intensive care clinicians, clinical geneticists and genetic counsellors. They explored every aspect of the testing process and how it could be made faster.

The rapids team provided genomic testing to 40 critically-ill children and babies. They were able to reduce the time to 16 days – almost 10 times faster than the standard. More than half of those children received a diagnosis, and a quarter had a change in care because of it.[6]

The study was so successful that its lead investigators received funding from Australian Genomics, the Medical Research Future Fund and philanthropic foundations to take it nationwide. They made further improvements – from specialist sampling machines to new analysis tools to a rapid review committee available seven days a week – bringing testing time down to just 2.9 days.[7]

**Rapid genomic testing gives doctors and families answers when they are desperately needed. Children can be spared more invasive tests, get treatments more likely to succeed, and go home to their families earlier.**

An application with the Independent Hospital and Aged Care Pricing Authority is underway for rapid testing of critically-ill children to be nationally funded. Meanwhile, Victorian and West Australian governments have stepped up to ensure that children in their states can access this test when needed.

6 "Meeting the challenges of implementing rapid genomic testing in acute paediatric care", Stark et.al, Genetics in Medicine (2018) doi:10.1038/gim.2018.37

7 "Integrated multi-omics for rapid rare disease diagnosis on a national scale", Lunke et.al., Nature Medicine (2022) **https://doi.org/10.1038/s41591-023-02401-9**

# 4 Genomics emerged as a powerful weapon in the fight against superbugs.

## Victorian hospitals now use genomic sequencing to stop superbug transmissions before they become full-scale outbreaks.

Antibiotic-resistant microbes ('superbugs') are among the world's most serious health threats. Arriving unseen in hospitals, they spread rapidly from one patient to another, causing longer admissions, spiralling hospital costs and even deaths.

A 2015 outbreak of drug-resistant *Klebsiella pneumoniae* affected patients in multiple hospitals across Victoria. Public health researchers began to wonder whether genomic sequencing could pinpoint which strain of a particular bug was circulating, and what treatment it might respond to.

A landmark Melbourne Genomics study, led by researchers from Austin Health and The Peter Doherty Institute for Infection and Immunity, sequenced the genomes of superbugs detected in multiple hospitals across the state.[8] By analysing genomic data alongside patient movement data, the hospital infection control teams could deduce exactly what type of bug was circulating, and how it was moving through the hospital – in time to quickly implement containment measures such as patient isolation and deep cleaning.

Lessons learnt from the study proved vital during the COVID-19 pandemic. The Doherty Institute's Molecular Diagnostic Unit led the public health laboratory response, enabling Victoria to start sequencing COVID-19 samples as early as March 2020 – launching one of the most rapid and comprehensive responses in the world.

A new superbugs study is underway. Having performed a 'genomic snapshot' of eight significant drug-resistant pathogens, the research team is now testing ways to speed up the collection of both genomic and patient movement data, and rapidly report this back to hospitals.

## Stopping superbug infections in their tracks will protect some of Victoria's most vulnerable patients.

8 "Key parameters for genomics-based real-time detection and tracking of multidrug-resistant bacteria: a systematic analysis", Gorrie et.al, The Lancet Microbe (2021) **https://doi.org/10.1016/S2666-5247(21)00149-X**

# 5 Victoria worked out how to safely store and share genomic data.

**Flash drives were once used to transport the blueprints of human beings – because laboratories lacked the capability to store and share them.**

One of the first challenges for the newly-formed Melbourne Genomics Health Alliance was to provide a platform for members to collaborate on genomic testing. While existing software tools could sequence and analyse genomic data, nothing on the market was capable of moving gargantuan genomic datasets between those tools.

Doctors, scientists, bioinformaticians and developers worked together for six years to build a software platform that could orchestrate a genomic test from beginning to end. Cloud computing was used at every step.

In 2019, laboratories began to use the platform – then called GenoVic – for live tests. New tools were integrated, and GenoVic was able to share data with other laboratory systems and even electronic medical records.

The next step was to enable laboratories to reanalyse data stored in the cloud, to find new answers for patients. Laboratories could even share data with each other, with strict governance processes in place to ensure patient consent and privacy.

By 2023, six laboratories had used GenoVic to run over 20,000 genomic tests and it was time to go nationwide. The platform was renamed *Genomical®* – a word first coined by researchers who recognised that 'astronomical' was insufficient to describe the big data generated by genomics.[9]

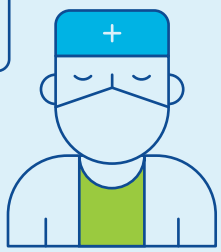
With Australia close to finalising a national approach to genomic information management, a platform like *Genomical* will be key to protecting Australians' genomic data and using it meaningfully in their healthcare.

**By 2023, six laboratories had used Genomical to run over**

## 20,000 genomic tests

9 'Big Data: Astronomical or Genomical?' Stephens et.al, PLOS Biology (2015) **https://doi.org/10.1371/journal.pbio.1002195**

# 6 Genomics is entering mainstream medicine.



**Once a highly niche field, genomics has become critical to multiple specialties: from paediatrics to nephrology, cancer care and even organ transplants.**

Back in 2013, even geneticists were just learning how to use genomics. Working on Melbourne Genomics clinical projects enabled them to bring this emerging knowledge into other areas of medicine.

Geneticists and diagnostic scientists were brought into multidisciplinary teams, working alongside oncologists, cardiologists, nephrologists or other medical specialists to diagnose patients and determine treatment options. This approach was critical to the high diagnostic rate of genomics, and became standard practice.

Today, clinical genetics services struggle to keep up with the demand for genomics. New models are being trialled to build genomics expertise within medical specialties, so that only complex cases are referred to clinical genetics.

Paediatricians are creating resources to streamline the ordering of Medicare-funded genomics tests. Nephrologists are leading multidisciplinary teams that meet to discuss patients who may need genomic testing. The teams include nephrologists, clinical geneticists, renal nurses and genetic counsellors.[10]

Oncologists are now trialling ways to access genomics expertise when their patients need it: from holding telehealth consultations with the Peter MacCallum Cancer Centre, to establishing 'genomics superusers' within cancer hospitals.

Meanwhile, genetic counsellors are joining the team at dementia and neurology clinics, to support families where dementia has a genetic origin. A groundbreaking project is underway to sequence the genomes of Victorians who will receive liver or kidney transplants, to prevent adverse reactions and increase the chance of success.

**Whatever the speciality, genomics is here to stay.**

10 See **paediatricgenomics.org.au** and **kidneygenomics.org.au**.

# More than 3,000 healthcare professionals have joined Victoria's genomic workforce.

## From geneticists to general practitioners, Victorian medical professionals are learning how to bring genomics into their field of expertise.

Building a genomics workforce in Victoria required both education and the opportunity to put it into practice.

The first priority was to ensure geneticists and laboratory scientists had the skills to perform genomic testing. Melbourne Genomics offered courses in variant interpretation (the detective work of identifying whether a particular gene variant can explain a specific health condition) and these were quickly filled.

Doctors needed to learn when to order genomic tests and how to use them in patient care. Melbourne Genomics projects provided hands-on experience, under the guidance of genetic experts. A genomics immersion program was also created, offering doctors six months of experience in genetics services, laboratories or research projects. Many of those doctors went on to become 'genomics champions' in their respective disciplines.[11]

Education programs were developed, with topics ranging from beginner (Genomics in the Clinic) to specialist (Genomics for Oncologists) to advanced (Clinical Variant Interpretation). Melbourne Genomics also teamed up with the VCCC Alliance to offer genomics micro-certifications to oncologists.

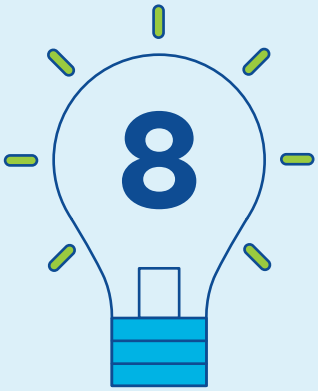
Between 2015-2023, more than 3,000 Victorian healthcare professionals learned how to bring genomics into their practice. An evaluation of these programs found that doctors gained not only knowledge, but the confidence to use it in healthcare.[12]

Victorian students can now choose several career paths in genomics. Among these are The University of Melbourne's Master of Genomics and Health and Master of Genetic Counselling, both of which feature content developed by Melbourne Genomics.

## Victoria now has the opportunity to cultivate a world-leading genomics workforce.

11 "'It's something I've committed to longer term': The impact of an immersion program for physicians on adoption of genomic medicine", Martyn et.al, Patient Education and Counselling (2020) **https://doi.org/10.1016/j.pec.2020.10.013**

12 Maher et. al. Genomics education for medical specialists: case-based specialty workshops and blended learning. Journal of Translational Genetics and Genomics. 2023; 7(2): 94-109. **http://dx.doi.org/10.20517/jtgg.2023.04**




# Alliance members are at the forefront of genomic innovation.

## Working together has opened doors to new facilities, powerful collaborations and future discoveries.

Victoria has become the go-to place for genomics researchers.

**WEHI** launched its Advanced Genomics Facility in 2021, enabling researchers to collaborate and access the latest genomic technologies. That same year, **The University of Melbourne** partnered with Illumina to create The Advanced Genomics Collaboration, aiming to lift local genomics research and clinical trials to a global scale and quality. The University also hosts Australian BioCommons, a national collaboration to build digital capability for life science research.

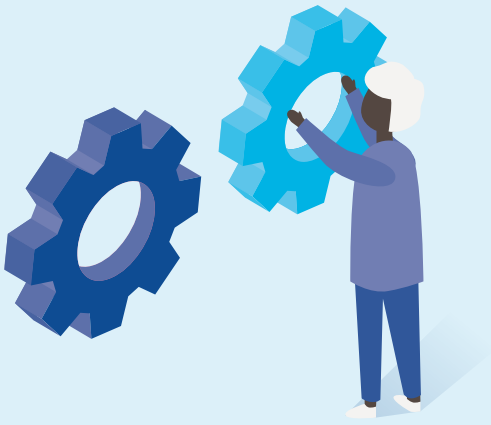
New partnerships are set to bridge the divide between research and clinical care. **Monash Health** partnered with the Hudson Institute of Medical Research to support the translation of genomics research into personalised treatments for cancer and inflammatory diseases. In July 2023, the **Peter MacCallum Cancer Centre** and The University of Melbourne announced a new centre to transform how precision oncology is delivered in Australia.

Collaboration is also making genomic testing more equitable and widely available. The pathology laboratories at all Alliance member hospitals have expanded the range of genomic tests they offer. Clinicians from **The Royal Melbourne Hospital** are trialling ways to use genomic reports to inform pre- and post-operative care of people receiving organ transplants. Funding from **The Royal Children's Hospital Foundation** helped expand the use of rapid genomic testing for critically-ill children, as well as the KidGen collaborative: where over 100 clinicians, researchers and scientists work together for people with genetic kidney disease. The **Murdoch Children's Research Institute** also played a critical role in these initiatives, as well as hosting Australian Genomics (a national genomics collaboration) and establishing the Centre for Population Genomics in collaboration with the Garvan Institute.

Meanwhile, **Austin Health** and the Doherty Institute are spearheading efforts to use genomic sequencing to control superbug transmission in Victorian hospitals.

## Alliance members are taking genomic technologies further than ever.

**CSIRO** is leading the development of data standards and cloud infrastructure to enable the safe sharing of genomic information. **AGRF** remain proud early adopters of state-of-the-art sequencing technologies and continue as a key partner in the Zero Childhood Cancer Program (ZERO), Australia's first national precision medicine program for children with cancer.



# 9 Community support for genomic medicine is growing.

**The last decade has seen a rise in the number of patients and families advocating for genomic medicine.**

Rare disease is surprisingly common. Around 8% of Australians live with one of more than 10,000 known rare diseases,[13] and their advocacy was critical to the formation of Melbourne Genomics.

The Genetic Support Network of Victoria (GSNV) had been active since 1997 as a voice for people with rare conditions. Rare Voices Australia was formed in 2012 as a national peak body, while Syndromes Without a Name (SWAN) Australia was founded in the same year by Heather Renton, whose daughter had a then-undiagnosed neurodevelopmental disorder.

Melbourne Genomics created a community advisory group (CAG) shortly after its own inception in 2013. Representatives from SWAN and GSNV joined as members, alongside other champions of consumer rights in healthcare. The CAG has advised on every project developed by Melbourne Genomics, ensuring patients and their families were considered at every step.

**Around 8% of Australians live with one of more than**

**10,000 known rare diseases**

Consumer voices have been essential in getting genomic tests funded. The Usher Syndrome community's advocacy helped shape the final decision on Medicare funding for infant hearing loss, while Maddie's Vision has helped genomics researchers gain funding for their work on bone marrow failure syndromes.

Meanwhile, families with genetic conditions ranging from Spinal Muscular Atrophy to Neimann-Pick Syndrome have connected over social media to form support groups. Their collective advocacy informed a national strategic plan for rare diseases, which was released in 2020. The rare disease sector continues to lead the charge for equitable, patient-centred genomic medicine.

Last year, First Nations genomics researchers created ALIGN: a national alliance to ensure that Aboriginal and Torres Strait Islander peoples benefit from genomic medicine. ALIGN privileges Indigenous leadership and sovereignty, and Melbourne Genomics is proud to support its work in Victoria.

13 Elliott, E & Zurynski, Y. (2015) 'Rare diseases are a 'common' problem for clinicians. Australian Family Physician. **https://www.racgp.org.au/afp/2015/september/rare-diseases-are-a-common-problem-for-clinicians**

# 10 There's an easier path between medical discovery and routine healthcare.

**Evidence suggests that it takes 17 years for research discoveries to enter clinical practice.[14] Victoria is showing how it can happen much faster.**

While the benefits of genomics were known a decade ago, doctors were a long way from being able to use this knowledge in their daily practice. Melbourne Genomics programs have become the gold standard for bringing innovation into healthcare.

Because healthcare is a complex adaptive system, Melbourne Genomics tackled multiple aspects of change simultaneously.[15] The real-world evidence for genomics was documented alongside the real-world barriers to its use. Health economists compared the cost of genomic medicine with the value it could deliver, while doctors and scientists found ways to make each step in the process quicker and more efficient. Clinicians were funded to bring genomics into their area of speciality, creating change leaders who have continued to take genomics forward.

Victoria has the right pieces in place to bring genomics into routine healthcare. It has evidence of when genomics is most relevant for Victorian patients. It has a world-leading data platform that makes genomic testing efficient and scalable. It has a broad cohort of clinicians and scientists who are skilled in genomics. And it will soon have frameworks and models of care to help health services provide genomic medicine safely and effectively.

**When the Melbourne Genomics program ends in 2025, Victoria will be well equipped to offer genomic medicine to all Victorians who need it.**

14 "It Takes an Average of 17 Years for Evidence to Change Practice-the Burgeoning Field of Implementation Science Seeks to Speed Things Up", Rubin, R., JAMA (2023) doi:10.1001/jama.2023.4387

15 "Preparing for genomic medicine: a real-world demonstration of health system change", Gaff et.al., NPJ Genomic Medicine (2017) doi:10.1038/s41525-017-0017-4

# For paediatric doctor Cara, understanding the cause of her son's kidney disease was crucial.

**I remember the obstetrician looking at my 20-week antenatal scan and going, "Whoo, those kidneys are bright!"**

After that, I had genetic testing via amniocentesis and regular ultrasounds for the rest of my pregnancy – but aside from bright spots on the kidneys, everything seemed normal.

Then Edward was born – and his Day Five renal scan showed multiple cysts on his kidneys. That news was really hard to hear, because I'd allowed myself a glimmer of hope. But knowing the cause of Edward's kidney disease was important, because that would give us an idea of his prognosis and what kind of care he'd need.

A/Prof Cathy Quinlan, a nephrologist at The Royal Children's Hospital, organised whole genome sequencing for Edward, me and my husband Steve. The test found two genetic variants in Edward: one known to cause cystic kidneys, and the other of unknown significance.

Genomic testing gave us a diagnosis of autosomal dominant polycystic kidney disease. It was actually the best diagnosis we could get – because it means Edward's kidneys aren't likely to fail during childhood. We also know that his disease-causing variant wasn't inherited, so our other son Henry is unlikely to have the same problems. Meanwhile, researchers are investigating the significance of the unknown variant.

Right now, Edward's living his best five-year-old life. He's a happy kid and full of beans. The diagnosis made it possible for us to stay on top of his condition with regular monitoring.

**As a paediatric doctor and as a parent, genomic testing has the ability to give us answers we really need.**

Photo: Rodney Dekker

# Melbourne Genomics 2013-2023

## 2003

- The first near-complete human genome is sequenced by the Human Genome Project.

## 2013

- Seven organisations (The Royal Melbourne Hospital, The Royal Children's Hospital, University of Melbourne, WEHI, Murdoch Children's Research Institute, CSIRO and AGRF) form the **Melbourne Genomics Health Alliance**. Their shared aim is to bring genomics into healthcare in Victoria.

## 2014

- **Five projects are launched** to investigate the clinical utility of genomics in real-world healthcare.
- Melbourne Genomics forms its Community Advisory Group.
- Experts plan for a **shared data management system** for genomic testing. This eventually becomes *Genomical*.
- Victorian Government funding enables twice as many patients to be offered genomic testing.

## 2015

- **First peer-reviewed paper from Melbourne Genomics projects published in *Genome Medicine*.**
- First education workshop for diagnostic scientists on **variant interpretation.**
- Victorian Government contributes $25M to Melbourne Genomics for a new four-year program.

## 2016

- **First clinical results from Melbourne Genomics projects published in *Genetics in Medicine*. They provide critical evidence for Medicare to fund genomic tests for some children.**
- Peter MacCallum Cancer Centre, Austin Health and Monash Health join Melbourne Genomics.
- Six new projects begin to investigate the use of genomics in **non-Hodgkin lymphoma**, **solid cancers**, **complex children's conditions**, **immunology**, **deafness** and **heart conditions**.
- First education event for clinicians. 105 people attend.
- Victorian Government commits $8.3M for public access to genomic testing for people with rare conditions.

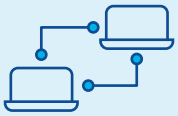
## 2017

- **The 1000th patient receives genomic testing through Melbourne Genomics program.**
- Five more projects investigate genomics in **bone marrow failure**, **neurological and neurodegenerative diseases**, **superbugs**, **kidney disease** and **perinatal autopsy**.
- Victoria releases its *Genetic and genomic healthcare 2021* framework.
- **Victorian Public Healthcare Award for Improving Children's Health** is jointly won by VCGS, Melbourne Genomics and The Royal Children's Hospital.
- Community Advisory Group recognised in the **Minister for Health Volunteer Awards**.

## 2018

- The first of 23 **evidence reports** is submitted to the Victorian Government – showing the medical benefits, cost-effectiveness and value of genomics.

## 2019



- **GenoVic**, a world-first software platform for clinical genomic data, goes live in Victoria.
- More than **3,000 patients** have now received genomic testing through Melbourne Genomics program.
- **1,000 people** have now participated in Melbourne Genomics education.
- Over 260 guests attend **'Translating Genomics, Transforming Care'** – a symposium to share results from Melbourne Genomics projects.

## 2020

- Medicare begins to fund genomic testing for children with suspected genetic conditions.
- Five laboratories are now using GenoVic for genomic testing.
- The Victorian Government announces $35M for a final Melbourne Genomics program.

## 2021

- A new **Controlling Superbugs** project begins. The project informs guidelines on using genomics to stop hospital superbugs, which are published in the *Lancet Microbe*.
- **10,000 clinical genomic tests** have now been run through GenoVic.

## 2022

- Medicare funds genomic testing for people with suspected genetic kidney disease and cardiomyopathy.
- New projects explore models of care for using genomics in **oncology**, **nephrology**, **transplants** and **dementia care**.
- The first 'genomic snapshot' of superbugs in Victoria is produced.

## 2023

- Healthcare leaders discuss the **implementation of genomics in Victorian hospitals**.
- Online resources help **paediatricians and nephrologists** to order Medicare-funded genomic tests.
- GenoVic software platform becomes ***Genomical®:*** *Your launchpad for genomic medicine*.
- Melbourne Genomics celebrates its 10th birthday.

## 2025

**The final Melbourne Genomics program comes to an end: equipping Victoria with tools, models and evidence to embed genomics in the health system.**

# Delivering value

**Genomics has delivered significant value to the Victorian health system, which will continue to grow as its use increases in routine healthcare.**

## $4.7M

Delivered to Victoria through use of Medicare-funded genomic tests

## >$150M

In research and government funding leveraging Melbourne Genomics member investment

## $20.8M

Estimated health system savings to date from use of genomic testing for conditions investigated by Melbourne Genomics*

*Based on evidence from our clinical projects.

## >$2.4M

Efficiency gains by using the Genomical software platform from 2021-23.

# Tess* underwent genomic testing to inform a potential kidney transplant.

## Nothing but a DNA test would have figured out what happened to me.

I'd had kidney stones for a long time. A little over three years ago, my urologist called me, saying: "You should see a nephrologist straight away." I could hear the panic in his voice.

My kidney's glomerular filtration rate had dropped to 12, which was considered 'end stage'.

The nephrologist put me on dialysis. I asked him, why am I on this path? He said: "Your kidney's not working. It's just you."

That wasn't good enough. I have two adult sons. If this was a 'just me' problem, couldn't it be just them, too?

I said, I want a DNA test. He said no, it wasn't at all necessary. I kept insisting. I needed to know, for myself and my sons.

Eventually I was sent to The Austin for my kidney transplant. There I met the doctor who turned out to be my saving grace. I said to him, is it too much to ask for my DNA to be tested? And he said, "Of course you can!" It was such a relief.

The DNA test found Primary Hyperoxaluria type 1. The genetic problem was in my liver, not my kidneys! Had I gotten a kidney transplant before this was discovered, the new kidney would have died because my liver would have been pumping it with excess oxalates.

I had just one operation in December 2022, to get a new kidney and liver. The results are good so far, and I'm about to go back to work.

My sons got a test, too. Both are carriers of the gene mutation that caused my illness, but it won't affect them.

**It really is important that doctors don't dismiss patients when they ask for a DNA test. It's not the right option for everyone, but at least talk it through. After all, I wouldn't be here today if I hadn't insisted on it.**

*Name changed to protect privacy.

**Melbourne Genomics**
Health Alliance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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

c/o WEHI
1G Royal Parade
Parkville VIC 3052

**melbournegenomics.org.au**

@ enquiries@melbournegenomics.org.au
Melbourne Genomics Health Alliance
@MelbGenomics


Melbourne Genomics acknowledges the Wurundjeri people of the Kulin Nation, on whose lands we work, and all Aboriginal and Torres Strait Islander 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.