Queensland Genomics

QUEENSLAND GENOMICS
JULY 2016 - JUNE 2021

Accelerating change in healthcare

Queensland Genomics was a $25 million Queensland Government investment over five years, to support projects and initiatives that bring genomics into everyday healthcare in Queensland.

Clinics and projects operating **in 7 Qld hospitals** caring for patients across Queensland

35+ genomics training and education events held

Launched a **Genomics Information Toolkit** for patients

Funded and supported **45** clinical, capability and community projects

Collaborated with **170+** researchers & clinicians

Sequenced **250+** patients with Myeloid cancer, with 70% being clinically relevant results

2000+ bacterium sequenced to track, trace and prevent infectious disease

Invested **$4.7 million** to uplift genomic testing capability in Queensland

Delivered a digital genomics 'Blueprint' for Australia guiding implementers and policy makers in the sharing of genomics information

Established **11** new nursing positions to embed genomics into mainstream Nursing and Midwifery practice

Established **4** multi-disciplinary Cardiac Genetics Clinics across the State, with 130+ referrals received across the clinics by May 2021

Developed an **Australian-first** test to detect cell fusion in Cancer

1300+ Patients recruited across Queensland into clinical projects

Delivered **'Genomic Partnerships'** - a set of Guidelines for genomic research with Aboriginal and Torres Strait Islander peoples of Queensland

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EXECUTIVE SUMMARY

The 'genomics revolution' continues to have a transformative impact on the delivery of health care, and this is well evidenced through renewed funding and prioritisation by government, industry, and consumer advocacy groups.

Advances in sequencing technologies means all of a person's genes can now be sequenced at once through genomic sequencing. This has enabled a growing capacity for accurate diagnosis and prognosis of disease, the development of targeted treatment strategies, and opportunities to assess predisposition to disease. So, beyond this disruptive technology, the question is 'how can we plan and invest to ensure equity, sustainability, privacy, and critical benefits to our population?'.

Our goal for the Queensland Genomics Program was to accelerate the adoption of genomics into everyday care for Queenslanders.

Genomic medicine has enormous potential, but with new technology comes new challenges. For Queensland, some of the challenges identified through Queensland Genomics' work included:

- poor genomic literacy among clinical and diagnostic workforces,
- limited methods for analysis of genomic sequences to a quality sufficient for patient care,
- inadequate systems, legal frameworks and policy to support the management of the huge volume of data generated by sequencing, and
- ensuring patient consent to genomic sequencing throughout the course of patient care.

Queensland Genomics was foremost a collaborative program and we worked with our partners to find solutions to many of the challenges brought about by this disruptive technology.

We worked to connect our health system and consumers with genomics research – taking purposeful and coordinated steps to fast-track the way to personalised medicine, and focussed on leveraging the strengths of our health system to meet these challenges.

Our approach

The program was designed to concurrently invest, over three funding rounds, in clinical activity, in building capability and capacity in our health system, and importantly, engaging our community to activate them in the discussion – putting people at the centre of their care.

In round 1, our program helped us discover the amazing genomics research potential in Queensland, and the strengths of our health system that we could draw on to advance our mission.

In round 2 we applied co-design and innovative investment approaches to make sure Queensland's genomics program established strong clinical leadership, effective and dynamic governance and well planned, achievable and measurable activity.

Finally in round 3 we empowered our health system to ensure more Queenslanders would benefit from genomics, and forward planning and investment could occur strategically and with greater certainty, navigating challenges and leveraging opportunity.

Throughout the program we considered workforce issues, health economics and sustainable funding, how to effectively build diagnostics capability here in Queensland, the ethical and legal implications of new technology like genomics, and considerations for our electronic medical record.

Via the Program, Queensland Health invested in a range of disease areas that were genomics ready – including cancer, neurology, cardiac and infectious disease. It is through these clinical pilot projects we were able to create capability in a systematic manner.

We worked with Health Consumers Queensland to establish a community advisory group and supported them to be active participants in the genomics program. Through the Queensland Genomics Community Advisory Group, we were able to meaningfully engage culturally diverse communities in genomic medicine, and develop and deliver genomics literacy education sessions in 2019 and 2020 to 213 multicultural health workers and interpreters.

Recognising the significant health disparity between Indigenous and non-Indigenous Australians, we also partnered with QIMR Berghofer Medical Research Institute on projects designed to support and engage

Above: Lindsay Fowles, Erin Evans, Katrina Cutler and David Bunker at a Queensland Genomics Community Advisory Group meeting in 2021.

Aboriginal and Torres Strait Islander peoples to access genomics and personalised medicine.

None of this could have been accomplished without the support and energy of the Queensland Genomics Business Team, the many collaborators and partners within the genomics community and the confidence and backing of Queensland Health. I would like to thank all of those who worked determinedly to bring people together and navigate through the many and varied challenges.

Closing this program has encouraged all of us to think about the future. It's clear genomics in health care must be guided by a clear policy and continued strategic investment. Queensland Health is developing this policy framework to help us make good decisions and guide the focus for precision healthcare in our state.

These next steps will mean the ideas and capability we've generated through the Queensland Genomics program paves the way for success in the future.

Queensland Genomics was foremost a collaborative program and we worked with our partners to find solutions to many of the challenges brought about through this disruptive technology.

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COMMUNITY ENGAGEMENT

Engagement projects led by the Queensland Genomics Community Advisory Group

The Queensland Genomics Community Advisory Group met quarterly for the life of Queensland Genomics, leading projects and providing input into the Queensland Genomics program. Projects included:

- Genomics Information Toolkit – genomics literacy materials for Queenslanders.
- Genomics Consumer Support and Advisory Group for Queenslanders – developed the scope for a consumer support service.
- Genomics Literacy in Multicultural Queensland – provided training in genomics to bilingual health workers and medical interpreters.
- Public education sessions – partnered with Health Consumers Queensland to build genomics literacy.
- Survey of Genomics Literacy in Queensland's Health Consumers – survey of health consumers.
- Community Advisory Group Overview Publication – built evidence for best practice consumer involvement.
- Mapping the Patient Journey – compared patient journeys across different jurisdictions.

Genomics Information Toolkit for Patients

The Genomics Information Toolkit is a resource to assist patients to engage with and access genetic and genomic health services and testing in Queensland.

The Toolkit was a suggestion from Gary Hondow from the Queensland Genomics Community Advisory Group, to give families accurate information about genetic testing, the different kinds of tests available, and where to go for genetic and genomic testing in Queensland.

Visit **queenslandgenomics.org/pct** to download the toolkit.

Queensland Genomics Community Advisory Group

The Queensland Genomics Community Advisory Group was a key part of the Alliance's collaborative model. The group brought together representatives from across the medical genomics community including patients, clinicians, researchers and health administrators.

MEMBERS

Dr Erin Evans (Chair), Dr Aideen McInerney Leo, Ms Louise Healy, Mr Greg Pratt, Ms Jessica Bean, Mr Gary Hondow, Dr Nic Waddell, Mr Satrio Nindyo Istiko, Dr Lindsay Fowles, Ms Deborah Robins, Ms Katrina Cutler, Mr David Bunker.

213

medical interpreters and bilingual health workers received accredited genomics literacy training

99,000+

health consumers reached through a digital campaign promoting a Genomics Information Toolkit

50

health consumers brought together to develop the scope for a Genomics Consumer Support and Advisory Group for Queenslanders

"I think one of the successes of the Queensland Genomics Community Advisory Group is that everybody felt that their voice was valued and everybody felt that they had something to contribute. They really felt a true collaboration in this group, and I think that is ideally what community advisory groups are trying to achieve. We are always trying to achieve co-design and collaboration, and I think we actually nailed it with this one."

DR ERIN EVANS

CHAIR, QUEENSLAND GENOMICS COMMUNITY ADVISORY GROUP

A Dr Lindsay Fowles, Genetic Counsellor, Queensland Genomics Community Advisory Group member.

B Queensland Genomics Community Advisory Group.

C Healthcare interpreters participating in *The Language of Genetics & Genomics* interpreter training session, 2019.

D Dr Sid Kaladharan, QIMR Berghofer Medical Research Institute, speaking at a Health Consumers Queensland genomics information session.

E Healthcare interpreters participating in *The Language of Genetics & Genomics* interpreter training session, 2019.

WHOLE OF LIFE

PROJECTS

- **Epilepsy:** Improving patient outcomes in neurological disorders: Piloting a neurogenetics service for refractory epilepsy patients.
- **Paediatric neurodevelopment:** supporting diagnostic access for rare neurodevelopmental and complex multisystem isorders across Queensland.
- **Whole genome sequencing in paediatrics:** Clinical whole genome sequencing in the diagnosis of paediatric genetic disorders, and acute care genomics rapid trio whole genome sequencing in neonatal and paediatric intensive care units.
- **Immunology:** Implementation of genomics into an integrated diagnostic and treatment service for primary immune deficiencies and other immune dysregulation syndromes in children.
- **Cardiac:** Queensland Cardiology Genomics Program.
- **Children's:** Genomics strategy for Queensland Children.

$3.65m

total investment into the whole of life portfolio

KEY HIGHLIGHTS

We have seen impressive outcomes across each of these projects.

- **Epilepsy:** This project has delivered insightful outcomes through next generation sequencing in epilepsy. The project has developed a Neuro-genomics clinic at Metro North HHS and successfully run post-testing multi-disciplinary team discussions.
- **Paediatric neurodevelopment:** This project has made genomic sequencing technology available to Health and Hospital Services throughout the state. The team have also built local clinical capability by creating a model of care for patients with rare neurodevelopmental and complex multisystem disorders in South-east and Northern Queensland. The project has improved the confidence of clinicians in identifying appropriate patients for genomic testing and understanding the referral process.
- **Whole genome sequencing in paediatrics:** This project led to the creation of the Illumina Partnership Program which brought together the Genomic Institute, Pathology Queensland and Illumina® working together to deliver a new standard of testing in paediatrics using whole genome sequencing. This project is implementing an integrated, clinically applicable diagnostic pipeline for whole genome sequencing in paediatric healthcare in Queensland and has set an ambitious recruitment target of 200.
- **Immunology:** This project implemented a clinical genetic diagnostic service to provide timely and accurate genetic diagnosis in severe primary immunodeficiency, immune dysregulation, auto inflammation and genetic bone marrow failure.
- **Cardiac:** The Queensland Cardiology Genomics Program has set up new multi-disciplinary Cardiology Genomics clinics across Queensland, including at the Royal Brisbane and Women's Hospital, The Prince Charles Hospital, Princess Alexandra Hospital and Cairns and Hinterland Hospital and Health Service. This has been achieved by face-to-face and telehealth clinics. The team has achieved equity in quality of care and supported continuity of care for patients being cared for by their local cardiac teams.
- **Children's:** The Genomics Strategy for Queensland Children will start in the second half of 2021; as part of this strategy, Children's Health Queensland will address equitable access to genomics for children throughout the State.

Taylor and Charlene Mundy: Visit **bit.ly/3il9TIi** to read more about their story ["The seizure just would not stop." One family's journey with Dravet Syndrome].

INFECTIOUS DISEASES

Genomic medicine is transforming the way we identify, treat and prevent infections.

Genomics stops superbug outbreak in special care unit.

Routine testing in 2018 in a Queensland hospital identified a number of babies in the hospital's neonatal unit tested positive to ESBL producing Klebsiella oxytoca, a multi-drug resistant and potentially virulent pathogen in neonates.

The usual approaches to tracking and preventing the bacteria from spreading weren't working, but by using genomic testing, the Infection Prevention and Control team managed to track it back to the source.

Initial genomic testing results suggested an environmental reservoir was the transmission source. Subsequent whole genome sequencing (WGS) was used to investigate 60 environmental sources of transmission during the outbreak.

The source of transmission was found to be detergent bottles used in the special care unit. No new cases emerged once these detergent bottles were removed. In this case, WGS was instrumental in revealing the route of transmission and guiding the infection control response. Controlling the outbreak early meant avoiding any serious harm to any babies.

This research was funded by Queensland Genomics and has been published in the ***Journal of Clinical Microbiology***.

The use of genomic testing in infection control could save 650 lives a year in Queensland.

A study by Queensland researchers found the routine use of genomic testing could prevent a significant number of hospital-acquired infections and related deaths every year.

Associate Professor Louisa Gordon, from QIMR Berghofer Medical Research Institute, led the study which assessed the impact of using whole genome sequencing (WGS) as part of routine surveillance in hospitals compared with standard microbiology testing over a five year period.

"We used WGS to check for six common bacteria which are resistant to multiple antibiotics and known to have serious consequences when hospital outbreaks occur," she said.

"We analysed the genomic sequencing data from clusters of these multidrug-resistant bacteria that emerged across 27 Queensland hospitals.

"Compared with standard care, we found using WGS in routine surveillance could help prevent 36,726 patients a year from being infected or colonised with the six most common multidrug-resistant bacteria. That could help prevent 650 associated deaths from bloodstream infections."

Researchers also analysed the costs to implement WGS in routine surveillance compared with standard of care.

"We found WGS would cost an additional $26.8 million a year to put into practice, however it would save $30.9 million a year due to a reduction in costs for cleaning, nursing, personal protective equipment, shorter hospital stays and antimicrobials," Associate Professor Gordon said.

The study was funded by Queensland Genomics and published in the journal ***BMJ Open***.

$2.08m

total investment into the
infectious diseases portfolio

PROJECTS

- **Hospital-acquired infections:** Using genomics to track, treat and prevent hospital acquired infections.
- **Sepsis:** Saving lives through the early detection of sepsis.
- **Sepsis North Queensland:** Tackling infections in remote communities.
- **COVID-19:** Preventing COVID-19 outbreaks in healthcare.

KEY HIGHLIGHTS

The infectious diseases genomics projects have delivered significant benefits to patients and the healthcare system.

- Implemented routine genomic surveillance and genomics outbreak response in Pathology Queensland.
- Supported management at the Princess Alexandra Hospital for OXA 181 outbreak.
- Enabled early precision detection of an outbreak at Caboolture Special Care Nursery.
- Established the Queensland bioinformatics pipeline for sepsis and microbial genomics
- Reduced the impact of an outbreak at the Royal Brisbane and Women's Hospital Burns Unit.
- Hosted a national Microbial and Infection Control Genomics Workshop in 2019.
- 2000+ bacterium sequenced.

Following significant investment, the clinical work is continuing either through MRFF or as embedded services within Pathology Queensland.

Paul Simmonds (left) and Hnin Aung (right) were part of the team at Pathology Queensland who developed the first NATA accredited test in Australia for haematological cancers.

PERSONALISED TREATMENT FOR PATIENTS WITH MYELOID BLOOD CANCERS

A pilot project in Queensland hospitals found genomic testing has improved the accuracy of diagnosis and treatment for patients with myeloid blood cancers, which affects the blood and bone marrow.

The project offers a more personalised diagnosis based on the individual patient, and the unique genetic makeup of their cancer.

Dr Cameron Curley, Director of Haematology and Bone Marrow Transplantation at the Royal Brisbane and Women's Hospital, said genetic information is key to selecting the best possible treatment for patients with myeloid cancer.

“In this pilot we have sequenced more than 250 patients who have been diagnosed with myeloid cancer, which is a common blood cancer affecting more than 3000 people in Australia every year,” he said.

“Data gathered through genomic sequencing has helped us understand how the cancer may behave in that patient, and how to best treat them.

“Myeloid cancers are difficult to treat, and a bone marrow transplant can cure some patients' cancer. The challenge for specialists is working out which patients will benefit from this intensive procedure.

“When using genomic sequencing in addition to standard testing, we found useful genomic variants in 70 per cent of patients. For 11 per cent of patients this information resulted in a change in their treatment plan.

“For example, we found some patients would benefit from a change to the timing for their bone marrow transplant.

“For others it meant they went on to receive a bone marrow transplant, whereas prior to genomic sequencing it was not indicated as necessary.

“For many patients, understanding their diagnosis and prognosis is almost as important as any therapeutic outcome. This pilot project provides many patients with accurate information on their diagnosis and what is likely to happen to them.”

“For 6 per cent of patients involved in the pilot, genomic sequencing identified possible hereditary links to myeloid cancer.

“Genomic data also identified some harmful gene mutations that the team had not previously been able to define. Acting quickly to treat patients in these circumstances has significant potential to prevent unnecessary progression of the disease and lead to better outcomes for patients.

"Over the next three to five years we will assess outcomes for patients' post-transplant, as well as longer term outcomes for non-transplant patients," he said.

CANCER

Over three funding rounds, Queensland Genomics invested in five cancer projects in Melanoma, Lung Cancer, Acute Myeloid Leukaemia, Acute lymphoblastic leukaemia and breast cancer. Investments were also made in the development of cancer nursing, and cancer genomics planning for adult and paediatric services.

$2.77m
total investment into the cancer portfolio

KEY HIGHLIGHTS

The Cancer Genomics Program has rapidly advanced Queensland capability in precision cancer care.

- Australian-first accredited test for fusion genes.
- Multi-disciplinary team meetings to support genomic testing for AML and ALL ensuring statewide equity of access and outcomes through the Qld Myeloid Genomics Program.
- Statewide overarching governance structure for cancer genomics implemented and led by Queensland cancer clinicians.

Through the introduction of a clinician led governance structure, Queensland is well placed to continue to implement genomics for cancer patients. It is a national leader for haematological cancer genomics in clinical practice and following initial investment from Queensland Genomics is leading Australia via MRFF for precision oncology for breast cancer patients.

PROJECTS

- **Queensland Myeloid Genomics Program:** Using genomic testing to improve the diagnosis, prognosis, and therapy options for patients with myeloid cancers.
- **Acute Leukaemia in Children:** Targeted genomic tests to provide rapid, comprehensive and cost-effective analysis of acute leukaemias, with a focus on children's leukaemias.
- **Queensland Implementation of Precision Oncology in Breast Cancer:** Using whole genome sequencing to identify the unique genetic profile of a person's breast cancer tumour.
- **Lung cancer:** Assessed the impact of genomic testing in lung cancer treatment in Queensland.
- **Melanoma:** Tested the use of genomics in the prevention and early detection of melanoma.

ABORIGINAL & TORRES STRAIT ISLANDER COMMUNITIES

In partnership with QIMR Berghofer Medical Research Institute, a series of projects were established to support and engage Aboriginal and Torres Strait Islander peoples in accessing genomics and personalised medicine.

PROJECTS

- **Genomic Partnerships** – guidelines for researchers partnering with Aboriginal and Torres Strait Islander peoples.
- **Indigenous Genomics Literacy Project** – culturally appropriate genomics information materials.
- **Genetic Health Pathways** – improving access to genetic testing and counselling services for Aboriginal and Torres Strait Islander peoples.

KEY HIGHLIGHTS

- Developed 'Genomic Partnerships: Guidelines for genomic research with Aboriginal and Torres Strait Islander peoples of Queensland'. You can download the guidelines at **bit.ly/GenetiQs.**
- Developed genomics education materials, which include brochures and a video, that are culturally appropriate and cover the topics of DNA, genes, genetic health, genetic testing and precision medicine. You can view or download these resources at **bit.ly/IG-HeLP.**
- Ran nine workshops across Queensland to discuss the genetic health needs of Aboriginal and Torres Strait Islander peoples, and issues in accessing services, in order to inform the referral pathway plan.
- Established a standardised approach to consent and the use of collected genomics information for research purposes.

ETHICS AND CONSENT

PROJECTS

- Ethics, Legal and Social Implications (ELSI)
- Statewide Consent and Ethics

KEY HIGHLIGHTS

- Developed a series of guidelines and policies to support community engagement, consent, research, justice.
- Established a standardised approach to consent, and using collected genomics information for research purposes.
- Significantly contributed to ELSI literature for genomics in healthcare including the publishing of peer-reviewed papers exploring ELSI issues for children, sharing of genomic data, public health, Australian and Queensland privacy legislation, Aboriginal and Torres Strait Islander people and communities, and diversity and inclusion for genomics medicine.
- Provided key recommendations to Queensland Health regarding the use of standardised consenting processes for genetics clinics utilising the Australian Genomics National Clinical Genomics Consent Forms.
- Undertook a patient survey of patients attending Genetic Health Queensland to examine patient opinions of genomic data sharing.

$530k

total investment

PATHOLOGY QUEENSLAND

Through the Queensland Genomics investment, Pathology Queensland has advanced its genomics capability from limited single gene testing and limited engagement with clinical services, to offering a full suite of genomics services.

Services now include a nation-leading gene fusion panel, myeloid panel, and whole exome and whole genome sequencing. Pathology Queensland now oversee a central sequencing fund for genomic sequencing in Queensland, and have established significant IT capability to enable integrated reporting, bioinformatics pipelines and clinical services.

$4.7m

total investment into
Pathology Queensland

1300+

patients sequenced within
7 clinical projects

2000+

microbial isolates
sequenced

INFORMATION MANAGEMENT SYSTEMS & DATA

Integrating clinical data with genomic data to improve coordination across multidisciplinary teams and drive more effective treatment for patients.

PROJECTS

- **Multidisciplinary teams data system** – Upgraded the existing Queensland Online Oncology Tool (QOOL) to support the Queensland Genomics Cancer and Epilepsy Projects.
- **Genomics Information Management** – Developed an architecture for the longitudinal management of Queensland genomic information, outlining the standards, policies and procedures required to support a common infrastructure for the safe, secure and privacy-centric management and use of genomic data to improve healthcare and support ethically approved translational research.
- **Genomics and ieMR** – Ensure Queensland's integrated electronic Medical Record (ieMR) supports genomic medicine through analysis and documentation of the integration steps required between the ieMR and other relevant technology systems to support the order entry – results reporting process. Ensuring clinicians can easily order genomic tests and access the results.
- **National Approach to Genomics Information Management** – Queensland Genomics led a collaborative project across Australia to develop a National Approach to Genomics Information Management. The project worked across all commonwealth, state and territory health agencies and with leading translational research programs and research infrastructure groups to build consensus on how to share data.

KEY HIGHLIGHTS

- **Developed the National Approach to Genomics Information Management Blueprint** - which sets out a series of principles to guide decision-making and future implementations, on the responsible collection, storage, use and management of genomic data.
- **Redevelopment of the Queensland Online Oncology Tool (QOOL)** - to include genomics services in haematological cancer, breast cancer and neurology.
- **Established the Statewide Genomics Digital Reference Group** - Run by the Genomic Institute, this group works to understand and enhance the practice of genomics within the digital world. This group has engagement from a range of stakeholders across the health system and works in collaboration to drive the digital agenda.

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WORKFORCE DEVELOPMENT AND EDUCATION

Knowledge Network

Conceptualised by Queensland Genomics and its workforce partners, and hosted by the Genomics Insitutute MNHHS, this project helped to build a genomics-ready workforce. This project worked across a number of disciplines to deliver workforce development and education resources.

The Genomic Institute Education Coordinator, funded by Queensland Genomics, has worked in the following areas:

- Renal Education: This project delivered five online modules in addition to decision aids for nephrologists. The decision aid will support Nephrologists in their referral for genomic testing.
- Maternal Fetal Medicine: This project will devise a Queensland specific education and training program for midwives and obstetric clinicians to better support patients and families.
- Primary Care (see 'Primary Care - Genomics for GPs section for more information).

Primary Care - Genomics for GPs

The Primary Care education project was run by CheckUP Australia, a non-profit organisation that works with GPs across the state. This project delivered three webinars to inform GPs about genomics and the referral process for genomic testing. Queensland Genomics funded resources at the Genomic Institute, and the project has now also delivered five online modules, which GPs can now access online.

Cancer Coordination

Sponsored by the Statewide Cancer Clinical Network, the project will shape the cancer genomics landscape in Queensland. Ultimately integrating cancer genomics services and pathways across the state.

Variant Curation Workshops

To further build core capabilities in genetic pathology within Queensland, Queensland Genomics partnered with Melbourne Genomics to deliver ‘hands on’ professional workshops on variant curation.

Nursing and Midwifery

Sponsored by the Chief Nurseing and Midwifery Officer, this initiative has worked with nurses and midwives from across the state to improve knowledge on genomics. Queensland Genomics has also funded 95 midwives to attend the Genetics in Pregnancy course at The University of Melbourne.

Masters in Diagnostic Genomics

Through a partnership with Queensland University of Technology, the project team established the first Masters in Diagnostic Genomics in Australasia. The new course at QUT is available for health professionals and researchers to enhance their knowledge of genomics. This course is supporting researchers, clinicians and laboratory scientists to develop essential knowledge and skills in genomics to gain accreditation as a diagnostic genomics scientist.

MinION Workshop

Delivered by experts from Oxford Nanopore in 2019, this workshop gave participants experience with real-time analysis tools as well as in-depth analysis workflows with command line tools.

Microbial & Infection Control Genomics Workshop

Held in 2019, this workshop demonstrated the potential of microbial genomics and how to apply it to hospital outbreaks and infection control management. The workshop was attended by 55 infection control practitioners and nurses, microbiologists, scientists and clinical researchers from across Australia.

A Professor Lyn Griffiths, project lead of the Masters in Diagnostic Genomics course, now available at Queensland University of Technology.

B Attendees at the Somatic Variant Curation Workshop 2019.

C The team at CheckUp Australia who were instrumental in delivering the Genomics for GPs education resources.

D Dr Natalie Thorne, Melbourne Genomics Health Alliance; Dr Ain Roesley, Peter MacCallum Cancer Centre. Presenters at the Somatic Variant Curation workshop 2019.

NURSING AND MIDWIFERY

Multidisciplinary teams have been essential for the successful implementation of genomics in Queensland. Nurses and midwives form an integral part of these teams and so Queensland Genomics, in partnership with the Office of the Chief Nursing and Midwifery Officer Queensland Health, worked together with other key partners to devise and deliver a comprehensive nursing and midwifery genomics program.

KEY HIGHLIGHTS

- Creation of 11 new genomics positions for nurses and midwives
- 106 nurses and midwives accessed in-depth genomics education via Nursing and Midwifery genomics education workshops delivered by QUT
- 95 midwives sponsored to attend Genetics and Pregnancy course
- Queensland Nursing and Midwifery Genomics Workforce Plan developed

$1.46m

total investment into nursing and midwifery

LESSONS LEARNT

STRATEGIC APPROACH

As a health innovation program Queensland Genomics' clarity of remit, goal and mission were essential to ensure a focused program and targeted investment for maximum impact. The underlying program remit was to support equity of access and high-quality care across Queensland. Key lessons from Queensland Genomics' strategic approach to the program include:

- Statewide impact – utilising statewide services has been key to sustainable investment
- Empower clinicians to lead – establishing clinical governance across multiple HHS's with strong links to established governance structures such as clinical networks.
- Meaningfully engage patient, families and the community – effectively achieved through a co-design and co-lead approach.
- Deliver for multiple partners - identifying partners' organisational goals and aligning investment to achieve these goals.

PROGRAM MANAGEMENT

Queensland Genomics developed a highly effectively project management methodology. Key lessons from Queensland Genomics' program management approach include:

- Project tracking - implementing project reporting that enables robust governance and transparency, but is not too resource intensive for busy clinicians, thereby supporting successful project delivery.
- Project delivery - Queensland Genomics Business Team attendance at Project Steering Committees (or other project wide meetings) enabled a nuanced understanding of project progress, risk and challenges, in a way that cannot be achieved via monthly reporting.
- Contracting and funding for outcomes and project progress - linking deliverables to payment milestones supports the achievement of projects.
- Resourcing - directly funding project officers or research assistants to manage the project is valuable and lessens the administrative burden on busy clinicians and researchers.

COMMUNITY AND CONSUMER ENGAGEMENT

Queensland Genomics' strong focus on community and consumer engagement enabled successes across the whole of the program. Key lessons from Queensland Genomics' community and consumer engagement approach include:

- Multidisciplinary teams - creating a community advisory group as a multi-disciplinary team including representatives from all sectors of the community (consumer, clinical, research, health administrator) built understanding across the genomics community.
- Supporting community-led projects – undertaking a co-design process with the community group to identify and build community-led projects results in project activities that respond to the needs of communtiy.
- Resourcing – providing a human resource to work with the community advisory group to help them to get things done and to build projects out of their ideas; and providing financial resources to pay consumers for their time, and external specialist suppliers to help to deliver on project activity.

OBJECTIVES MET

CLINICAL	COMMUNITY	CAPABILITY	OVERALL PROGRAM
1. Workforce able to incorporate genomics into healthcare Achievement status: Sustained advancement			
Queensland Genomics has worked across a number of clinical areas including cancer, pathology, nursing and midwifery, general practice, paediatrics, infectious diseases, infection prevention and control, cardiology, epilepsy/neurology, renal, medical interpreters and consumers. However, genomics has the potential to impact on many more clinical areas into the future, by strategic partnerships and investment Queensland is well placed to continue to advance workforce confidence, knowledge and capability in genomics.	The Queensland Genomics community group targeted specific areas of the workforce such as medical interpreters, and Aboriginal and Torres Strait Islander community controlled services (in partnership with QIMR Berghofer and Queensland Aboriginal and Islander Health Council).	In collaboration with specific clinical areas Queensland Genomics has built a strong workforce development capability. This has seen: investment at the Genomic Institute for an Education Coordinator resulting in education modules for renal, baseline genomics and other areas. The investment has also seen the development of a Nursing and Midwifery Workforce Plan and Education resources for GPs. The establishment of legacy partnerships both within and external to Queensland Health, will continue to support the development of priorities for an ongoing workforce plan via the Queensland Health Genomics Executive Working Group.	Due to the restructure of the Queensland Genomics into a clinical program with embedded clinical leadership, the program has been able to progress workforce development in several key areas. This, combined with a focus on community and overall capability in workforce development, has resulted in a sustained advancement for the Queensland workforce to incorporate genomics into healthcare.
2. An evidence base for clinical genomics Achievement status: Sustained advancement			
Queensland Genomics has worked across the breadth and the depth of many specialty areas. In the paediatrics space Queensland now has a much stronger understanding of the evidence base for, not only testing, but clinical service delivery. We also have a deeper understanding within certain sub-specialities, such as paediatric immunology, intellectual disability, and Acute lymphoblastic leukaemia in paediatrics. This same scenario plays out in cancer, where we now understand genomics for haematology as well as somatic, germline and solid tumours. This deep analysis and understanding has allowed Queensland clinicians and Pathology Queensland to build a precision approach for patients groups and a broad approach for specialty areas.	The Queensland Genomics evidence base has now expanded beyond clinical engagement and we have incorporated patient and community opinion and feedback into the evidence. This has resulted in a much more holistic and rich data set to inform decision making.	Queensland Genomics' fully integrated approach to evidence includes not only evidence derived from results but also clinical and community opinion and experience. We have also established several mechanisms for the gathering and sharing of the multiple levels of evidence and data including the operationalising of genomics MDTs, where the evidence is constantly reviewed in the clinical setting. The value of the data createad by the Queensland Genomics investment, extends beyond test results, with scientist, treating clinicians and referring clinicians coming to a precision diagnosis and care pathway, using all available evidence.	The shift away from evidence for research to implementation into clinical practice has enabled evidence to drive policy development, service planning, and ongoing investment decisions for Queensland Health.
3. Timely and cost-effective diagnostic workflows Achievement status: Sustained advancement			
The introduction of genomics into the statewide pathology service has not been without challenges; timeliness continues to be an issue due to workforce constraints. However, the investment has allowed Queensland to understand these issues and through the Queensland Genomics program Pathology Queensland has established valued partnerships with clinical teams and external laboratories to ensure clinical service delivery is not impacted by these issues. Instead Pathology Queensland has been able to double workforce capacity, triple test output and support MDTs across Queensland for diagnostic workflows.	With a focus on a staged approach to scalability and statewide access, the community of Queensland will be able to access equitable and timely testing that considers the impact of the constrained fiscal environment of health services in Queensland.	Pathology Queensland had extensive systems in place for cost-effective service delivery, but lacked an investment avenue to incorporate new testing approaches. Queensland Genomics' investment provided the opportunity for Pathology Queensland to become a national leader in genomic testing and put in place a plan for business-as-usual service delivery beyond Queensland Genomics.	Investing in the existing strengths of the Queensland Health system has built the development of timely and cost effective diagnostic workflows in the statewide pathology service provider, Pathology Queensland. Early recognition of the Australian focus on importance of pathology services via accreditation processes from the Royal College of Australasian Pathologists and NATA ensured Queensland was aligned to this national approach and meant that investment in testing resulted in NATA accredited results that met clinical and health system need.
4. Public awareness and understanding Achievement status: Significant advancement.			
The adoption of genomics in clinical practice and the implications for patients varies depending on the type of test, t clinical service, and patient need. The Queensland Genomics program has been able to, in several clinical areas, build an understanding of the complexity and support clinicians to address these issues whilst developing their clinical practice in genomics.	Significant progress has been made to co-design with consumers and patients, relevant education materials for patients and the broader community. This includes the development of a Patient Genomics Information Toolkit which was created by the Queensland Community Advisory Group and has been distributed across Australia. This important resource will be housed by Genetic Health Queensland post 30 June 2021.	Through the embedding of co-design across the Queensland Genomics program, we have ensured that multiple partners and stakeholders have been involved in efforts for public awareness and understanding, resulting in residual capability across Queensland to support ongoing awareness of genomics in the community.	The emergence of genomics as a clinical tool for patient care will continue to evolve. Queensland Genomics have been able to create awareness around the appropriateness and applicability for patients and the community and have left a legacy that should support any further re-iterations as the field continues to advance.

CLINICAL	COMMUNITY	CAPABILITY	OVERALL PROGRAM
5. Genomic sequence results are used for the benefit of patients Achievement status: Sustained advancement in specific clinical areas.			
Test results across a number of clinical specialities are now integrated into patient care. The clinical and patient benefit has been realised in anumber of ways including improved diagnosis and prognosis, elimination of potential diagnoses (even if a definitive diagnosis wasn't found), and improved and rapid access to appropriate care and treatment.	With the focus on patient benefit we have seen tests support patients in a number of areas including entering remission earlier than anticipated due to a precision diagnosis. Clinicians are also anticipating higher survival rates for AML patients following rapid access to bone marrow transplantation. We have also seen children enter the right care pathway significantly earlier and avoid catastrophic conditions due to delays in diagnosis. In cardiac, we have seen families get answers to genetic concerns and have supported patients to access appropriate screening programs. We have prevented or stopped infectious disease outbreaks in Queensland. In cases where we don't have direct patient benefit due to the structuring of the project we now understand the reasons behind this. For example, in our breast cancer project, we understand concerns that women have in entering into a genomic research program, such as the potential of genomics delaying fertility treatment for women who want to have families. This insight means that into the future Queensland can potentially amend the structure of testing processes and clinical pathways to ensure that genomics seamlessly integrates with the goals of patients.	Acknowledging the importance of clinical benefit and system benefit has meant that Queensland has only invested in sequencing that is integrated into clinical practice (either as an implementation project or an innovation project) but also that this sequencing doesn't just benefit patients but also supports clinicians and the broader health care system.	The shift from research to clinical implementation means that Queensland Genomics projects are no longer answering research questions but are resolving real-time clinical concerns and questsions.
6. A system for the management of clinical genomic data Achievement status: Advancement			
There is now a deep understanding of the value of genomic data within clinical services via Pathology Queensland. Queensland Genomics adheres to NATA accreditation requirements for genomic clinical services with results captured in AUSLAB and where possible, available through the ieMR.	Community representative groups were engaged in the development of the National Approach to Genomics Information Management project and the resulting Blueprint. The project worked extensively with Aboriginal and Torres Strait Islander groups to establish a set of guiding principles for the use and re-use of genomics data. Community Advisory Groups from Queensland Genomics, Melbourne Genomics and Australian Genomics were engaged to provide input to and validate the Blueprint's principles for consumer and Community principles.	Significant investment has occurred to support the capability development for the management of clinical genomic data. This has culminated in the development of the National Approach to Genomic Information Management (NAGIM) via Queensland Genomics and Queensland Health. The NAGIM project produced two key outputs, the NAGIM Report (which provided recommendations to the Project Reference Group), and the NAGIM Blueprint (which has been released to the public to support implementers working in jurisdictional health agencies and research groups across Australia). Under the NAGIM Blueprint, Australian Genomics will be convening national stakeholders and commence infrastructure prototyping, to deliver a series of phased recommendations, based on evaluations of research and clinical solutions.	The nature of digital transformation and the broader digital advancement program in Queensland has meant that Queensland Genomics has had to utilise existing systems for the management of genomic data. However, there are several plans developed that will greatly assist should investment and opportunity emerge for new systems to be implemented.
7. Accelerated translation of research Achievement status: Significant advancement			
By recognising that applicable research exists to support Queensland health needs, Queensland Genomics has been able to work with clinicians to find the right research solution for translation of research into clinical practice. This has enabled fit for purpose research translation.	Not applicable	Queensland Genomics has designed a translation assessment system which sees co-design and strategy alignment as key functional elements of decision making processes. This embedded system now involves multiple stakeholders who understand the system and can utilise this approach again should the need arise.	The foundations are now in place to accelerate research and translate it into everyday healthcare across a number of clinical specialties. Queensland Genomics has proved that research translation can occur rapidly and effectively. However, traditional approaches to research granting and research program development cannot be applied. Instead, Queensland Genomics developed and implemented an innovative investment program that saw the articulation of health need matched with research and clinical capability to deliver state-wide capability and clinical services.
8. A positive contribution, nationally and internationally Achievement status: Significant advancement			
The clinical program has demonstrated how genomics adoption can occur across a state population. Queensland Genomics collaborators have shown how genomics can be implemented into health systems and through their important roles in national and international groups and their experiences in the Queensland Genomics program, they can continue to positively contribute to the discourse for genomics implementation.	The Queensland Genomics Community Advisory Group was an innovative approach to consumer and community engagement. This has been acknowledged nationally as a leading program and an exemplar for how to undertake meaningful and purposeful engagement.	The Queensland Genomics projects have supported the development of 40 publications within the genomics national and international community and this evidence will be a valuable resource for genomics programs worldwide.	Following the governance review in 2018 Queensland Genomics diverted from the approaches of other Genomics programs in Australia. The benefit of this divergence is only now becoming apparent, as Queensland is well advanced in delivering genomic medicine across the state. Furthermore, due to the program, Queensland is also well positioned to progress precision medicine with plans underway within Queensland Health to maximise the opportunities created by Queensland Genomics.

PUBLICATIONS

ROUND 1 CAPABILITY BUILDING WORKSTREAMS

Ethics, Legal and Social Implications

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ROUND 1 CLINICAL DEMONSTRATION PROJECTS

Infectious Disease Clinical Project

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ROUND 2 CAPABILITY PROJECTS

Evaluation of Clinical Genomics

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ROUND 2 CLINICAL PROJECTS

Infectious Disease Clinical Project

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Community Organisations

CheckUp Australia
Epilepsy Queensland
Ethnic Communities Council of Queensland
Health Consumers Queensland
Queensland Aboriginal and Islander Health Council (QAIHC)
Queensland Fertility Group

Other Genomics Health Alliances

Australian Genomics Health Alliance
Melbourne Genomics Health Alliance