Media release

3 November 2016

Results demonstrate the success of genomic medicine

Patients get quicker, more accurate diagnosis and more personalised care when genomic sequencing is delivered within healthcare, an Australian-first study shows.

Victorian Health Minister, The Hon Jill Hennessy, today announced key findings from the Demonstration Project of the Melbourne Genomics Health Alliance, which during 2014-2015 provided genomic sequencing to selected Victorian hospital patients and evaluated its usefulness for medical practice.

We found that when doctors provide genomic sequencing to patients within Victoria's healthcare system:

- at least 6 times more patients receive a diagnosis
- patients receive care tailored to their individual genetic make-up
  (28% of children and 21% of all diagnosed patients had a change in care)
- patients can have fewer tests because genomic data can be stored and analysed again and again
  (one in 10 undiagnosed patients received a result this way, a number expected to increase as we learn more about the genome from researchers)

Importantly, the study provides evidence that, for certain medical conditions, genomic sequencing can replace other tests – resulting in better use of precious healthcare dollars.

Key findings regarding patients were:

- Patients want genomic sequencing (more than 90% of those approached agreed to the test)
- 96% of patients said they had enough information to make a decision about testing after genetic counselling
- Patients want their data stored for future analysis and research
  (98% agreed to this for uses relating to their condition, and 93% agreed to share data for any research)

"This work shows when and how genomic sequencing can bring benefit to patients, doctors and researchers," said Melbourne Genomics' inaugural Board Chair, Catherine Walter. "For infants with genetic syndromes, the results are spectacular: sequencing yielded 5 times more diagnoses at 75% less cost per diagnosis. That means fewer invasive medical procedures for these children."

"For undiagnosed children, having access to genomic sequencing early on and as part of mainstream healthcare will give the best chance of diagnosis and better treatment plans," said Heather Renton, President of SWAN Australia, an advocacy group for undiagnosed families. "It can save years of anxiety from not knowing. If families are able to share data for further research, this can only be a good thing, as more children will be diagnosed."

The Demonstration Project provided genomic sequencing to 315 patients, with one of five medical conditions, through a highly collaborative alliance of leading healthcare and research organisations in Melbourne.

Genomic sequencing resulted in a diagnosis for almost half (42%) of the adults and children with hereditary neuropathy (inherited forms of muscle weakness), whereas none had received a diagnosis through usual testing.

Doctors changed medical care for at least one-fifth of diagnosed patients, due to more precise diagnosis of their condition (28% of children and 21% overall). Changes included stopping unnecessary medications and check-ups, and providing new treatment and surveillance for early detection of known complications.

"In the next three years, Melbourne Genomics Health Alliance will further evaluate the use of genomic sequencing for 11 more medical conditions," said Catherine Walter. "Our collaborative and comprehensive approach to bringing genomics into healthcare is now also being implemented through the Australian Genomics Health Alliance and the Queensland Genomics Health Alliance – gathering more evidence for how we can best use genomics in medicine."

Media background

3 November 2016

About the Demonstration Project

The Demonstration Project was developed and funded by the seven founding members of the Melbourne Genomics Health Alliance, to investigate how and when genomic sequencing can improve healthcare.

The founding members of the Melbourne Genomics Health Alliance are:

- The Royal Melbourne Hospital
- The Royal Children's Hospital
- The University of Melbourne
- The Walter and Eliza Hall Institute of Medical Research
- The Murdoch Children's Research Institute
- CSIRO
- The Australian Genome Research Facility

The Demonstration Project was officially launched in 2014 and ran for two years. The Victorian Government supported the project's second year, with an allocation of funding that doubled the number of patients tested.

The Demonstration Project provided genomic sequencing to 315 patients, with one of five medical conditions:

- Hereditary neuropathy (inherited forms of muscle weakness)
- Focal epilepsy (seizures affecting one side of the brain with no known structural or environmental cause)
- Hereditary colorectal cancer (inherited syndromes causing colorectal cancer)
- Acute myeloid leukaemia (a cancer of the blood)
- Childhood syndromes (children with features suggestive of a single gene disorder)

The Demonstration Project has:

- determined the benefits to patients of genomic sequencing in comparison to tests usually provided for their condition, and
- developed a pathway for doctors to provide sequencing to patients within Victoria's healthcare system.

A Community Advisory Group was established at the outset, to give input across the work program and ensure consideration of community values, perspectives and priorities.

The Demonstration Project collected data on the views and experiences of patients undergoing genomic sequencing, as well as from clinicians and scientists on how genomics impacted their practice.

About Melbourne Genomics Health Alliance

We are an alliance of leading healthcare and research organisations dedicated to bringing the global knowledge of genomics to benefit the individual care of Victorians.

Genomic medicine holds huge promise for human health, but, across the world, experience shows that there are many challenges to implementing genomics into healthcare.

These include:

- knowing which patients will benefit;
- building skills and knowledge among the healthcare and other professionals involved;
- developing secure and ethical approaches for managing the 'big data' generated by genomic sequencing; and
- bringing the latest genome research to benefit patients sooner.

The Alliance's comprehensive program of work tackles these inter-linked challenges: assessing genomics in practice, establishing the best systems, harnessing the latest research, building healthworkers' skills and knowledge, and ensuring appropriate access to quality information.

Melbourne Genomics' vision is for Victoria to be a world leader in the use of genomics in healthcare.

This alliance of Victoria's healthcare and medical research strengths, supported by government, is uniquely placed to develop the enormous potential of genomics to improve our health.

Media background

3 November 2016

About genomics

Genomics has huge potential to improve healthcare. Looking at a patient's genome may help doctors diagnose illness more quickly and easily, as well as working out what medical care might best help.

Genomics is a rapidly advancing field worldwide, which examines an individual's complete set of genetic information (the genome) to identify changes (variants) that may impact on health.

The genome is your complete set of DNA, including all of your genes. Each gene has a specific function. When a gene contains a variation or change, it may not work properly and this may affect your health.

A genetic test looks at one or a small number of genes at a time. A genomic sequencing test can capture information from all genes at the same time, enabling scientists to analyse all of the genes relevant to a patient's condition in the one test.

Genomic sequencing can also be performed on cancer tissue, to look for changes in the tissue that may have implications for treatment.

How are gene changes (variants) identified?

Computer programs are used to analyse the large amounts of raw genomic data generated by sequencing of a person's genome. The computer scans this data for differences or 'variants' between the genetic sequence of the person being tested and a reference sequence.

This list of variants is then examined to see if any can explain the medical condition someone might have, or (in the case of cancer) identify the most appropriate treatment.

The first step is to establish whether a variant or change in a gene will stop it working properly. To do this, a medical scientist examines large databases and scientific papers to identify what is known about the relevant variants.

Then a team of experts – including medical laboratory scientists, bioinformaticians (statistical specialists who deal with biological data), specialist genetics doctors, genetic counsellors and other medical specialists – match the list of variants which may impair gene function against a person's health and family medical history.

This is how we can determine if a particular variant is likely to be the underlying cause of a genetic disease. Or, in other cases, whether a variant in a cancer genome may affect how a person might respond to particular treatments.