Genetic and genomic healthcare for Victoria 2021
Improving the health and wellbeing of Victorians
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## Acknowledgments

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To receive this publication in an accessible format, email <genetics@dhhs.vic.gov.au>

Except where otherwise indicated, the images in this publication show models and illustrative settings only, and do not necessarily depict actual services, facilities or recipients of services.

Where the term ‘Aboriginal is used, it refers to both Aboriginal and Torres Strait Islander people.

Indigenous is retained where it is part of the title of a report, program or quotation.

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Available at <www.health.vic.gov.au/genetics>
Genomics has huge potential to improve the diagnosis and treatment of illness in people of all ages.

As we increasingly use technology that can quickly and cheaply sequence DNA, we better understand the cause of medical conditions. And once we understand the ‘why’, we can then work out how best to treat or manage those conditions.

Genetic testing has historically played a key role in identifying some rare medical conditions in children and during pregnancy.

Now, genomics can be used to provide information that can be used to better manage many other common medical conditions affecting adults, like cancer and heart disease.

We no longer have to take a ‘one size fits all’ approach to healthcare. Genomics provides the information needed to deliver more personalised treatment and management of medical conditions.

Clinical DNA testing can make an accurate clinical diagnosis, identify cancers that respond to chemotherapy, identify personal risk to potentially adverse drug reactions and improve donor matching in bone marrow and organ transplantation.

Genomics can even help identify opportunities to prevent illness such as in families with a high occurrence of breast cancer.

Genome sequencing is not restricted to humans. It can also be used to identify microorganisms and detect antimicrobial resistance, which can better target antibiotic use thereby reducing the potential to develop antibiotic-resistant ‘superbugs’.

Preventing and controlling infectious disease is an important public health activity. In public health, genome sequencing quickly identifies infectious disease outbreaks through improved surveillance and can also aid vaccine development.

Genomics knowledge is changing almost daily.

As genomics is increasingly incorporated into routine healthcare and public health, we need to make sure clinical evidence supports the appropriate use of genomic information to improve the health of all Victorians.

We need a workforce trained and confident in using genomic information to improve patient outcomes.

We also need to make sure that Victorians are comfortable with how their personal genomic information is stored and used, and ensure it is not used to their disadvantage.

Genomics is increasingly playing a vital role in how we deliver healthcare in Victoria.

The Victorian Government’s Health 2040: Advancing health, access and care strategy (November 2016) sets the vision for healthcare in Victoria.

This document is about what needs to be done to ensure genomics contributes to that vision.

Hon. Jill Hennessy MP
Minister for Health
Minister for Ambulance Services
Executive summary

It has been more than 60 years since the discovery of the structure of deoxyribonucleic acid (DNA) and over a decade since the first DNA mapping of all the genes in a human (that is, sequencing of a human genome) was completed.

Since then, there have been major advances in technology. Along with increasing community expectations, these are bringing about a rapid increase in the pace at which information about genomes is being discovered, gathered, analysed and applied. This brings both challenges and opportunities for healthcare delivery, public health infectious disease surveillance and biomedical research in Victoria.

It is important to ensure that Victoria has, and continues to have, a sound basis for healthcare provision and is well placed to take advantage of new healthcare knowledge and approaches.

*Health 2040: Advancing health, access and care* states that most Victorians enjoy a high standard of health and wellbeing, but good health is not shared evenly across the population. In addition, demand for healthcare is rapidly growing and changing, and how healthcare is provided needs to change to keep Victorians healthy into the future.

Information from a health reform summit and public submissions was that:

- individuals, carers and the workforce need a greater say in how services work
- higher levels of chronic medical conditions require different thinking about health and healthcare
- not all healthcare is of high quality
- some groups of Victorians are more likely to experience medical conditions
- innovation could do more to prevent medical conditions and improve care.

Additional views on the future use of information from genomic sequencing (genomic information) in healthcare were collected through expert advisory groups, a workshop and public submissions provided in response to a discussion paper.

What Victorians thought was:

- There needs to be better understanding of the benefits and limitations of genomic information in the general population, among individuals and their families seeking healthcare and among healthcare professionals.
- There are questions about the current capacity and capability of health services to use genomic information.
- There are challenges in handling genomic information because of the large amount of data and the need to protect personal privacy.
- Individuals and their families want to be able to benefit from genomic information faster but to know that it is safe to use the information.
- There are some actions best taken nationally rather than just within Victoria.
- There may be opportunities to use genomic information to improve treatment and to prevent individuals getting sick.
- There are some uses of genomic information that Victorians need to decide are appropriate.
The vision in *Health 2040: Advancing health, access and care* is built around three goals:

- better health – promoting health and wellbeing
- better access – fair, timely and easier access to care
- better care – world-class healthcare every time.

Using genomic information in routine healthcare can help meet the goals in *Health 2040: Advancing health, access and care* through:

- faster and more accurate diagnosis of medical conditions
- better targeted treatment and prevention of medical conditions
- improved monitoring of the health of Victorians.

Health services in Victoria are already changing to become more focused on individuals and to provide safer, higher quality services.

Including genomic information into routine healthcare requires additional work such as:

- strengthening the healthcare system so that Victorians, regardless of their age, location or background, benefit from safe, fast and fair inclusion of genomic information into routine healthcare
- building trust so that Victorians are confident they are being provided with the best possible care and that their and their family’s genomic information will be handled and used in accordance with their wishes
- raising awareness about the use of genomic information in healthcare, its benefits and limitations
- growing knowledge so that Victoria is a leader in using genomic information in routine healthcare.

This can be done by the Victorian Government, other organisations and governments (state and Commonwealth), health workers, individuals, families and communities all working together.

Four priorities identified by Victorians for action in the next 12 to 24 months are:

- developing and implementing a statewide genetic and genomic services plan to ensure more equitable access to appropriate and sustainable services
- establishing a genomic health clinical network to improve the safe and fair adoption of genomic healthcare practice by the health workforce
- undertaking community consultations to address some of the key ethical, legal and social issues associated with including genomic information into routine healthcare to inform Victorian Government policy and funding decisions
- reducing superbugs and improving detection of infectious disease outbreaks through strengthening of microbial genomics activities in Victoria to improve the health of Victorians.

The Department of Health and Human Services will report annually on progress in delivering these and future priorities.
Introduction

‘Every so often, a scientific advance offers new opportunities for making real advances in medical care … We believe that the sequencing of the human genome, and the knowledge and the technological advances that accompanied this landmark achievement, represent such an advance.’

(House of Lords Science and Technology Committee, Genomic Medicine, Vol I: Report, 2009)

It has been more than 60 years since the discovery of the structure of deoxyribonucleic acid (DNA) and over a decade since the first DNA mapping of all the genes in a human (that is, sequencing of a human genome) was completed.

Since then, there have been major advances in technology. Along with increasing community expectations, these are bringing about a rapid increase in the pace at which information about genomes is being discovered, gathered, analysed and applied. This brings both challenges and opportunities for healthcare delivery, public health infectious disease surveillance and biomedical research in Victoria.

It is important to ensure that Victoria has, and continues to have, a sound basis for healthcare provision and is well placed to take advantage of new healthcare knowledge and approaches. Victoria’s health service providers, public health reference laboratories, universities and medical research institutes need to be supported to lead future discovery and innovation. In addition, the health service system needs to be prepared to apply new knowledge safely and fairly to improve the health and wellbeing of Victorians.
In healthcare, genetic information is currently used for many different purposes including to:

- diagnose a medical condition
- assess the risk that an individual will inherit a medical condition
- assess the health of a developing fetus
- understand the makeup of a cancer and find the best treatment
- select the correct blood, tissue or organ for matching across individuals
- identify and characterise a harmful micro-organism, such as a bacterium or virus, and the spread of antibiotic resistant micro-organisms.

Prior to genome sequencing, most genetic testing involved reading the DNA of a single gene. The time taken to read the DNA and report the result varied with the size of the gene. Testing was usually focused on the gene, or genes, thought to cause the medical condition.

Laboratory technology (genome sequencing) now exists that can read, in order, the DNA molecules of all the genes in an individual (in other words, the machines can sequence genomes) quickly and cheaply in one test. For humans, all 24,000 genes can be sequenced in a single laboratory test in less than a week.

Information from genome sequencing is being used to:

- better understand what causes medical conditions
- better understand why different people react differently to the same medical condition or treatment
- find differences and similarities among medical conditions
- more accurately identify and characterise micro-organisms
- identify the most effective medical treatment for each individual.

Including genomic information in a person’s everyday healthcare has the potential to reduce medical conditions, improve recovery from medical conditions and reduce the cost of providing healthcare.

Faster identification of micro-organisms means that outbreaks of infectious disease and the source of the outbreak can be identified quicker. This means fewer people become ill and fewer businesses are affected. New vaccines may also be developed, based on genome sequencing information, to prevent the spread of infection.

In addition, rapid identification of micro-organisms helps target antibiotic use in individuals. Timely and safe use of antibiotics also reduces the chance of micro-organisms becoming resistant to antibiotics.

More information is being discovered about genomes, medical conditions and genome sequencing laboratory technology. This provides new opportunities for Victorian researchers and companies to grow and is why the Victorian Government has identified genomics as a priority area in its:

- *Healthier lives, stronger economy: Victoria’s health and medical research strategy 2016–20*
- *Victoria’s international health strategy 2016–2020*
- *Medical technologies and pharmaceuticals sector strategy.*
Victorian healthcare now

Human genetic services

Victoria has specialist genetic clinical and laboratory services that provide diagnosis and risk assessment of genetic conditions (including cancer) in babies, children and adults and in pregnancy. Demand for services has grown by more than 50 per cent over the last five years (see Figure 1).

Figure 1: Number of public clinical consultations 2011-12 to 2015-16

In addition, genetic information is used in blood and tissue typing, organ donation and transplantation and in choosing drugs for cancer treatment.

Referral to a specialist clinical genetic service is usually made by a health practitioner. There are very few private genetic services in Victoria because very few genetic tests are funded through Medicare. About 75 per cent of medically ordered genetic tests provided in Victoria are funded by the Victorian Government.

The number and complexity of clinical genetic tests has grown steeply over the past few years, increasing spending on tests and creating confusion for doctors about what tests are useful and are free to the patient.

Microbial diagnostic services

Preventing and controlling infectious disease is important because there are often serious consequences for individuals, communities and the broader economy. Access to timely and accurate information about the type of micro-organism involved and the number of cases of an infectious disease is critical in recognising an outbreak, finding the source and acting to reduce the further spread of infection.

Many technical methods are currently used to identify and describe micro-organisms and Victoria’s public health reference laboratories are critical for finding and identifying harmful micro-organisms. Genome sequencing is changing how testing is performed and the information that is available from testing.
Research and its translation into healthcare

Use of genome sequencing technology has highlighted issues in how to analyse, report and store large amounts of information (often referred to as ‘big data’), workforce roles and responsibilities and health service system capacity (both clinical and laboratory). The Victorian Government has provided $25 million in funding to build genome sequencing capability in Victoria.

Alliances are being established across organisations locally, nationally and internationally, with many Victorian agencies, researchers, clinicians, ethicists and public health professionals involved in gene discovery and determining how genomic information is, and should be, used. These people and agencies are working together and sharing their knowledge and skills to build the evidence needed to improve Victorian healthcare.

Other research is occurring in the health and medical research sector, with health service providers, universities, medical research institutes and industry also working to improve healthcare in Victoria.
Why healthcare needs to change

*Health 2040: Advancing health, access and care* states that most Victorians enjoy a high standard of health and wellbeing, but good health is not shared evenly across the population. In addition, demand for healthcare is rapidly growing and changing, and how healthcare is provided needs to change to keep Victorians healthy into the future.

Information from a health reform summit and public submissions was that:

- Individuals, carers and the workforce need a greater say in how services work.
- Higher levels of chronic medical conditions require different thinking about health and healthcare.
- Not all healthcare is of high quality.
- Some groups of Victorians are more likely to experience medical conditions.
- Innovation could do more to prevent medical conditions and improve care.

Additional views on the future use of genomic information in healthcare were collected through expert advisory groups, a workshop and public submissions provided in response to a discussion paper.

What Victorians thought was:

- There needs to be better understanding of the benefits and limitations of genomic information in the general population, among individuals and their families seeking healthcare and among healthcare professionals.
- There are questions about the current capacity and capability of health services to use genomic information.
- There are challenges in handling genomic information because of the large amount of data and the need to protect personal privacy.
- Individuals and their families want to be able to benefit from genomic information faster but to know that it is safe to use the information.
- There are some actions best taken nationally rather than just within Victoria.
- There may be opportunities to use genomic information to improve treatment and to prevent individuals getting sick.
- There are some uses of genomic information that Victorians need to decide are appropriate.
Victorian healthcare by 2021

By 2021, Victorians could expect that:

- There is statewide access to safe genomic healthcare based on individual need.
- Individuals needing genomic healthcare are able to access the services they need earlier and regardless of their age, location or background.
- Genetic and genomic service delivery is based on a clear statewide service plan.
- Genomic services are provided by a competent health workforce confident in providing quality genomic healthcare.
- Genetic and genomic information is incorporated into the patient’s standard medical record.
- Micro-organisms are detected and identified using genome sequencing.
- Clear guidelines are used to decide when genome sequencing may be funded by the Victorian Government as part of routine healthcare.
- The Australian Law Reform Commission will have reviewed and updated its 2003 report on how best to handle personal genetic and genomic information.
- Trusted information on genome sequencing is available on the Better Health Channel website.
- Victoria has a genomics network that provides independent advice about genomic healthcare in Victoria.
- Victoria is a national leader in, and continues to contribute nationally and internationally to, the growth of knowledge about using genomic information in routine healthcare.
- Victoria uses its genomic expertise to grow investment in local research, innovation and jobs.
Examples of the health benefits of genomic sequencing

Sequencing fetal DNA that is circulating in a pregnant woman's blood may help detect a genetic condition in the fetus she is carrying. This may avoid the risk of miscarriage from using a needle to remove cells directly from the placenta for DNA testing, which is current practice.

Different cancers may have similar DNA profiles. This means that drugs developed to treat one type of cancer could potentially be used to treat other cancer types as long as they have a similar DNA profile. This may speed up access to existing specialist cancer drugs.

In 2015 a superbug infected several patients in a Victorian hospital at different times. Genomic sequencing identified the source of the superbug. Without sequencing, the connection between the patients would not have been made nor the source of infection confirmed. This led to a targeted response, thus preventing infection of other patients.

The Melbourne Genomics Health Alliance has found that six times more children with a suspected genetic condition are likely to receive a diagnosis if genomic sequencing is used rather than current genetic tests. This discovery reduces the likelihood of children being subjected to unnecessary, and costly, hospital testing and ineffective treatments over a number of years in attempts to find and treat the cause of their symptoms.
A new strategy for Victoria

Victoria’s health system can be a world leader in illness prevention, healthcare innovation and clinical care. People should be able to access healthcare regardless of their age, location or background. The vision in Health 2040: Advancing health, access and care is built around three goals:

• better health – promoting health and wellbeing
• better access – fair, timely and easier access to care
• better care – world-class healthcare every time.

Other strategies that support genetic and genomic healthcare in Victoria include:

• Victorian public health and wellbeing plan 2015–2019
• Victorian cancer plan: Improving cancer outcomes for all Victorians
• Koolin Balit: Victorian Government strategic directions for Aboriginal health 2012–2022

Growing genomic knowledge and building associated economic benefits will occur through:

• Healthier lives, stronger economy: Victoria’s health and medical research strategy 2016–20
• Victoria’s international health strategy 2016–2020
• Medical technologies and pharmaceuticals sector strategy.

In addition, Victoria’s genetic and genomic healthcare strategy will complement the National Health Genomics Policy Framework.

Genomic healthcare in 2021

Based on feedback received, Victorians would like to see genome sequencing used to improve their health and wellbeing. To do that, information about an individual’s genome needs to be used, along with other personal information, to identify the risk of a future medical condition occurring and, where possible, to lower that risk, to accurately diagnose a medical condition and to provide the best available treatment, including avoiding drug reactions.

The health workforce needs to be competent, and confident, in using genomic information to improve healthcare for Victorians. In addition, the workforce needs to be supported by information systems that assist in managing and using the genomic information to improve individual outcomes.

With improved risk assessment, early intervention (treatment before the medical condition gets too severe) and illness prevention (where a medical condition is avoided altogether), fewer people may fall ill. However, if they do, they would be more likely to get the right treatment the first time. This means that health services may cost less to provide and may be more readily available for people who need care.

In addition, couples could have the opportunity to know the chance of having a child affected by a genetic condition. They would have the chance to prepare earlier for their child’s special needs or consider other ways to grow their family without passing on a genetic condition.
There could be fewer micro-organisms resistant to antibiotics because micro-organisms will be identified faster and the right antibiotic given to the individual from the start of treatment.

Fewer people may get sick from food poisoning because outbreaks and their sources are found quicker.

People expect new treatments and interventions for medical conditions to be available quickly and at no or a low cost. Because new information about genomes, medical conditions and treatment is being found all the time, this information could be made available in Victorian health services quickly, safely and fairly.

Genomic information is very personal, and individuals need to be confident that their information is secure, handled in accordance with their wishes and not used to their disadvantage.

Finally, Victoria could be a world leader in genomic research and in the safe and rapid inclusion of genomic information into routine healthcare.

**Opportunities**

Using genomic information in routine healthcare can help meet Victorian health goals through:

- faster and more accurate diagnosis of medical conditions
- better targeted treatment, prevention and follow-up of medical conditions
- improved monitoring of the health of Victorians.

For Victorians to benefit from information about genomes in their healthcare, further work is needed to make sure that:

- people are able to access health services regardless of where they live in Victoria
- the use of genome sequencing improves people's health and reduces healthcare costs
- the benefits of research are transferred to routine healthcare quickly, safely and fairly.

**Getting there**

Health services in Victoria are already changing to become more focused on individuals and to provide safer, higher quality services.

Including genomic information into routine healthcare requires additional work such as:

- **strengthening the healthcare system** so that Victorians, regardless of their age, location or background, benefit from safe, fast and fair inclusion of genomic information into routine healthcare
- **building trust** so that Victorians are confident they are being provided with the best possible care and that their and their family's genomic information will be handled and used in accordance with their wishes
- **raising awareness** about the use of genomic information in healthcare, its benefits and limitations
- **growing knowledge** so that Victoria is a leader in using genomic information in routine healthcare.

This work needs to be done by the Victorian Government, other organisations and governments (state and Commonwealth), health workers, individuals, families and communities all working together.
Strengthening the healthcare system

Goals
- Victorians benefit from the safe and fair use of genomic information regardless of their age, location or background.
- Victorians have access to the right health service at the right time to meet their needs.
- Genomic information is included in routine healthcare quickly, safely and fairly.
- Victoria has a competent health workforce confident in using genomic information to benefit individual and public health.

Achievements
The Victorian Government has:
- Created genetic service ‘hubs’ in selected metropolitan hospitals that also provide services to other hospitals in metropolitan, rural and regional locations.
- Improved funding and access to familial cancer centres through the Victorian cancer plan 2016–2020.
- Established statewide services for infants, children and adults with inborn errors of metabolism (such as phenylketonuria) in age-appropriate public hospitals.
- Released:
  - *Health 2040: Advancing health, access and care* to improve Victorians’ health and wellbeing
  - *Digitising health: How information and communications technology will enable person-centred health and wellbeing within Victoria* to assist Victorians to benefit from information technology.
- Allocated $8.3 million in 2017–18 to support public funding of gene sequencing for paediatric and adult rare diseases and undiagnosed condition, a first in Australia.

Opportunities
- Increase the capacity and capability of health services to safely and fairly include genomic information in routine healthcare.
This will be realised through the Victoria Government:
- working with health workforce training bodies to ensure that:
  - genomics is included in undergraduate training for future clinical and laboratory positions
  - genomics education is available as part of postgraduate and continuing professional development training for the existing health workforce
  - training positions in genomic healthcare are available
- working with local, interstate and national bodies to address the immediate need for trained bioinformaticians to assist in the analysis, management and understanding of genomic information
- developing a statewide genetic and genomic services plan that covers clinics, laboratories and health information technology and ensures that access to genomic healthcare is driven by need regardless of an individual’s age, location or background
- making sure that genomic healthcare is respectful of the needs of different cultures
- aligning Victoria’s activities in health information and health information technology with national and international standards and agreements
- establishing a clinical network to assist in improving health workforce genomic knowledge and reducing variation in healthcare practice
- developing a plan for integrating pathogen genomics into the state-funded health system
- exploring how best to use genomic information to reduce medical conditions.
Building trust

Goals

- Victorians are confident they are being provided with safe and fair genomic healthcare.
- Victorians are confident their personal genomic information is handled and used as they want.

Achievements

The Victorian Government has:

- Funded the Melbourne Genomics Health Alliance to ask people how their genomic information should be handled.
- Funded the Genetic Support Network of Victoria to provide information and advocacy on behalf of Victorians with genetic conditions.
- Funded the University of Melbourne to undertake a project on the ethical, legal and social issues regarding genomic information.
- Submitted a Victorian Government submission to the Senate Standing Committee on Community Affairs Inquiry into Gene Patents.

Opportunities

- Have a discussion with Victorians about how genomic information could be used in healthcare.

This will be realised through the Victoria Government:

- assessing the value of genome sequencing in routine healthcare and surveillance of infectious diseases
- developing guidelines to help the Victorian Government decide when genomic information could be safely and fairly included in healthcare in Victoria
- developing community engagement strategies to better understand Victorians' views on the use of genomic information in healthcare, including the handling of unrelated or unknown test results
- working with other Australian governments and the Australian Law Reform Commission to update its 2003 report *Essentially yours: The protection of human genetic information in Australia*. 
Goals

• Victorians are aware of the benefits, risks and limitations of genome sequencing and genomic information.

Achievements

The Victorian Government has

• Funded the Cancer Council Victoria to provide information on heritable cancers.
• Funded the Genetic Support Network of Victoria and the Thalassaemia and Sickle Cell Society of Australia to provide education to the public on inherited medical conditions.
• Funded the Melbourne Genomics Health Alliance to hold public events on genomic healthcare.

Opportunities

• Raise awareness in Victorians about the role of genetics and genomic information in their health and wellbeing and protecting public health.

This will be realised through the Victoria Government:

• exploring the role of family history information collection in raising the awareness of medical conditions in families
• reviewing, updating and expanding information on genome sequencing on the Department of Health and Human Services’ Better Health Channel website
• working with other Australian governments and organisations to develop consistent, easily understandable and culturally sensitive information on genetics and genome sequencing
• developing strategies to increase Victorians’ understanding of the safe and fair use of genomic information – for example, through undertaking community consultations.
Growing knowledge

Goals

- Victoria is a leader in including genomic information in healthcare quickly, safely and fairly.
- Victoria adds to international genomic information and knowledge for both clinical and research use.

Achievements

The Victorian Government has:

- Provided funding of $25 million to the Melbourne Genomics Health Alliance to build genome sequencing capability in Victoria.
- Launched the following to support health and medical research, clinical trial access and growth in the biotechnology sector:
  - Healthier lives, stronger economy: Victoria’s health and medical research strategy 2016–20
  - Victoria’s international health strategy 2016–2020
  - Medical technologies and pharmaceuticals sector strategy.
- Driven healthcare innovation through funding the Better Care Victoria Innovation Fund, the Victorian Cancer Agency and clinical networks.
- Participated in the development of the National health genomics policy framework.

Opportunities

- Be clear when it is safe and fair to include genomic information in routine healthcare.
- Continue to contribute to the global knowledge of using genome sequencing to improve human health and wellbeing.

This will be realised through the Victoria Government:

- creating a genomics network to drive safe and fair inclusion of genomic information into routine healthcare
- developing guidelines to help the Victorian Government decide when and if genome sequencing is ready for inclusion in routine healthcare
- working with other governments and research bodies to grow knowledge about using genomic information in routine healthcare
- assisting Victorian laboratories as national leaders in the area of infectious disease genomics.
Next steps

There are a number of actions outlined in this document that will mean Victorians benefit from the use of genomic information in routine healthcare and public health.

Four priorities have been identified by Victorians for Victorian Government action in the next 12 to 24 months. They are:

- **developing and implementing a statewide genetic and genomic services plan** to ensure more equitable access to appropriate and sustainable services
- **establishing a genomic health clinical network** to improve the safe and fair adoption of genomic healthcare practice by the health workforce
- **undertaking community consultations** to consider some of the ethical, legal and social issues associated with incorporating genomic information into routine healthcare (such as health literacy and secondary findings).
- **reducing superbugs** and improving detection of infectious disease outbreaks through strengthening of microbial genomics activities in Victoria to improve the health of Victorians.

Other actions described in this document will build on these activities.

The Department of Health and Human Services will report annually on progress in delivering these priorities and future actions at <www.health.vic.gov.au>. 