**August 2016**



**Genomic Health Care for Victoria**

**A DISCUSSION PAPER**

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# Introduction

It has been over 60 years since the discovery of the double helix structure of deoxyribonucleic acid (DNA) and more than a decade since the completion of the first phase of the Human Genome[[1]](#footnote-1) Project.

Since then, there have been significant advances that, coupled with increasing community expectation and increasing community health literacy, are bringing about a rapid increase in the pace at which genomic information is being discovered, gathered, analysed and applied. This poses both challenges and opportunities for health care delivery, public health surveillance and biomedical research in Victoria.

It is important to ensure that Victoria’s public health system is, and continues to be, supported by a sound platform and is well placed to take advantage of new knowledge and approaches. Victoria’s health services, public health reference laboratories, universities and medical research institutes need to be positioned to lead future discovery and innovation and the service system needs to be capable and well prepared to apply this new knowledge safely and ethically in order to improve the health and wellbeing of all Victorians.

## Document purpose

Victoria’s last Genetics Services Strategy was published in 2005. It is time for a new strategy that provides a framework guiding the appropriate service and infrastructure planning of public genetic and genomic healthcare in Victoria into the future.

This discussion paper is part of the process for developing a new Victorian genetic and genomic services strategy. Its contents are aligned with broad Victorian Government policy directions, including *Health 2040: A discussion paper on the future of healthcare in Victoria* and the communique released following the Victorian Health Reform Summit, which identified key principles to guide future health reform.

This discussion paper includes input already provided from both internal and external expert advisory groups established to inform genetic and genomic strategy development. Vignettes have been included to illustrate aspects of genetic and genomic healthcare.

Public feedback on this discussion paper will be used to shape the new strategy and inform future development of a statewide genetic and genomic service and infrastructure plan.

# Background

## Human genetic services

Victoria’s genetic services comprise specialist clinical and laboratory services that support and inform reproductive, paediatric and adult diagnosis, risk assessment and management of heritable medical conditions. Genetic services have expanded significantly since the 1960s, when the original focus was on paediatric and reproductive medicine, to now include *inter alia* applications such as:

* Prenatal, such as non-invasive prenatal testing
* Adult, such as cancer and heart disease
* Safety and prevention (pathogen genomics, pharmacogenomics)
* Treatment (pharmacotherapy) and
* Organ donation and transplantation.

Waiting times for public genetic clinic consultations are currently up to nine months from initial referral, resulting in anxiety and potentially lost opportunity for early intervention for individuals and families who are waiting for a consult, and frustration for clinicians. Urgent access can be facilitated if clinically necessary, noting this usually requires travel to Melbourne where clinic frequency is higher than in regional and rural Victoria.

The current ineligibility of genetic counsellors to access Medicare rebates and the absence of private health insurance coverage of genetic testing means there are limited private clinical genetic services available in Victoria since patients usually incur out of pocket costs, including for genetic tests that might be ordered. Some medical specialists in other fields may order genetic testing and general practitioners may order some genetic tests; however, fewer than two dozen of the more than 4,000 genetic tests available are Medicare-funded. Some research projects include access to genetic testing and counselling that is covered by project funding.

The number and complexity of clinical genetic tests has increased steeply over the last few years, resulting in increased cost and creating confusion for health professionals about what tests are funded and for what indications.

Clinical DNA testing is now available to not only establish or confirm a clinical diagnosis but to also characterise individual genomes and tumours to inform treatment, identify predispositions to potentially adverse therapeutic reactions and improve donor matching in stem cell and organ transplantation.

Each clinical discipline is being challenged on how it should appropriately integrate genomic knowledge into current practice. Each is devising innovative multidisciplinary approaches to clinical care. However, to provide most benefit, the challenge now is to broaden the current *ad hoc* approach into a systematic and clinically appropriate incorporation of genomic knowledge into the public healthcare setting, including clinical, laboratory, research and education.

## Microbial diagnostic services

Preventing and controlling infectious disease outbreaks is an important public health activity as there are often social, financial and economic consequences to the individual, communities and the broader economy. Access to timely and accurate information indicating an increase in the number and type of cases of a disease or condition of public health concern is critical in recognising an outbreak, identifying the source of the infection and commencing necessary action to reduce further possible transmission and minimise negative impacts.

Victoria’s public health reference laboratories are a critical service provider in the state’s surveillance activities for identifying and confirming the presence and characteristics of pathogenic microbes of public health importance. An array of laboratory-based microbiological assays is utilised to identify and characterise pathogenic organisms. However, while traditional assay methods have been sufficient to date, the advancement of rapid, low-cost genomic sequencing technologies is transforming public health globally.

In addition, rapid identification and genetic characterisation of micro-organisms facilitates better targeting of appropriate antibiotic use. While not only improving treatment responsiveness, genomic sequencing also reduces the potential for development of antibiotic-resistant strains.

The impact of microbial genetics and genomics on healthcare can be summarised as follows[[2]](#footnote-2):

|  |  |  |
| --- | --- | --- |
| **Activity** | **Patient care** | **Public health** |
| Pathogen identification | Diagnosis | Outbreak detection |
| Antibiotic selection | Proper treatment | Effective antibiotic use |
| Vaccine development | Better protection | Reduced burden of disease |

## Genomic sequencing

The advent of massively parallel DNA sequencing, where multiple genes or even whole genomes may be sequenced rapidly in one test, has opened up opportunities for quicker and more accurate diagnosis, better targeted (precision) treatment and rapid identification and characterisation of pathogenic organisms. It has also highlighted issues in data management (‘big data’), workforce roles and responsibilities and health service system capacity (clinical and laboratory).

Alliances are being established locally, nationally and internationally, with many Victorian agencies, researchers, clinicians and public health practitioners actively involved in discovery, translation and sharing of genomic information.

In parallel, genomic sequencing that provides information on topics such as ancestry and wellbeing is being marketed directly to the community. This ‘consumer-driven genomics’ raises issues not only about the quality of information provided but also about the management of personal information, both of which have ramifications for relatives, future offspring and personal privacy.

**Vignette 1**

A fit and healthy 17 year old boy died while swimming. Post mortem examination could not reveal a cause of death. DNA sequencing identified a mutation in the KCNQ1 gene responsible for Long QT syndrome. His parents were both tested, which found that his mother was a carrier of this mutation. His three siblings were also tested and his older brother was found to have the same mutation; he was subsequently referred to a cardiologist for ongoing management and surveillance. The two younger sisters did not have the mutation and were released from cardiac screening. The extended family also underwent predictive testing. The genomic investigation not only answered the question of this tragic loss, but facilitated predictive testing and a management plan for those at risk, as well as release from screening and treatment for those not considered at risk.

# Development of a new strategy

The *Genetics Services Strategy for Victoria 2005 – 2009* set principles and directions to guide planning, development and delivery of Victoria’s public genetic services over that period. Since then, Victoria has operated without an overarching vision that appropriately harnesses new genomic sequencing knowledge and technology. In March 2015 an internal departmental reference group and an external expert advisory committee were formed to flag issues and develop concepts and scope for a Victorian genomic health care strategy. These groups met several times, culminating in a joint workshop in August 2015[[3]](#footnote-3). The following are key findings from the workshop.

## Summary of key findings from 2015 workshop

* The rapid expansion and translation of genomic technologies into clinical practice in the absence of clear clinical, legal and ethical frameworks requires immediate attention.
* There is low health literacy in both the community and among health professionals regarding the genetic basis and implications for many common and debilitating conditions, impacting knowledge uptake and use.
* Access to genetic care is not equitable across Victoria. There should be equal access for equal need.
* There is increasing demand for somatic/tumour testing to inform cancer treatment and patient management, including participation in clinical trials.
* Diagnostic odysseys are common for those individuals and families with rare and complex disorders. A good family history will improve the recognition of potential heritable disorders for these individuals and families and may reduce the burden and distress of the current diagnostic odyssey by reducing the time to definitive diagnosis. The importance of taking a good family history cannot be underestimated.
* Genomic healthcare poses many challenges for the health workforce. There is a need for: new skills and knowledge for health professionals to assist individuals and families dealing with complex information; better structures for working across disciplines to appropriately incorporate genomic information into clinical decision making; improved ways to ensure scarce resources are used efficiently; and development of a bioinformatics workforce to support research and translation into routine clinical care.
* The world of biomedicine is rapidly changing, creating a deluge of data that is placing significant pressure on current systems and approaches to processing and analysing information. A more robust and sustainable platform is needed to support bioinformatics and information sharing locally, nationally and internationally, with clear ‘rules’ about the use and sharing of such data.
* Victoria has some of the best academic health centres and research institutes in the country. There is a strong desire to leverage current investment to become a lead innovator in this space.
* The genetics technology landscape is evolving rapidly. Clinicians and researchers will benefit from clear guidelines and frameworks to support innovation, best clinical practice and world leading research.
* Microbial sequencing is rapidly replacing traditional laboratory methods used to identify and characterise pathogens.

**Vignette 2**

A 27 year old man diagnosed as HIV positive has therapy that includes the drug Abacavir, which can cause serious adverse reactions in some patients; this is linked to a particular immune system gene, HLA B\*57:01, which reacts with the drug metabolites. Pre-screening patients for the HLA B\*57:01 has eliminated the risk of adverse reaction and enables the appropriate therapy to be prescribed.

# Key factors

A number of already identified factors likely to impact genetic and genomic healthcare provision over the next ten years can be broadly grouped as:

* Models of care and service system capacity and capability
* Literacy – public, health professionals
* Personalised (precision) medicine including personalised prevention[[4]](#footnote-4)
* Co-productive healthcare[[5]](#footnote-5)
* Ethical, legal and social issues associated with genetic and genomic information
* Value for money – healthcare needs to be safe, high quality, evidence-based, clinically effective and cost effective.

These factors are expected to profoundly impact genetic and genomic healthcare provision over the next ten years.

**Vignette 3**

Between 2014 and 2015, increasing numbers of the critical antibiotic-resistant bacteria carbapenem-producing *Klebsiella pneumoniae* (CPE) were referred to the Molecular Diagnostic Unit Public Health Laboratory (MDU PHL) in Victoria. It was unclear if these cases were being imported from overseas or if the pathogen was spreading within Victorian hospitals. Using whole genome sequencing, MDU PHL was able to demonstrate a number of genomic transmission networks, which were then completely validated using detailed epidemiological investigation. The use of highly specialised long-read sequencing was also able to demonstrate the transfer of resistance elements between bacterial species. Combined whole genome sequencing analysis and epidemiological investigation is now applied to all new cases of CPE in Victoria. This work demonstrated the power of genomic sequencing to reveal transmission networks of drug-resistant bacteria within the hospital environment and between facilities, resulting in targeted interventions to curtail spread of the pathogen.

***Q1. Are there other factors/issues likely to impact genetic and genomic healthcare that should be considered? If so, what are they?***

# A new strategy for Victoria

The Victorian Government believes Victoria’s health system can be a world leader in prevention, innovation and clinical care, and should be accessible to people regardless of age, location or background.

The Department of Health and Human Services is committed to delivering on the Government’s vision and has identified the following core pillars underpinning a patient-focussed and innovative healthcare system:

* A strong focus on person-centred services
* Local solutions
* Earlier and more connected support for people
* Advancing quality, safety and innovation.

Within this broad framework, the following statements have been proposed to prompt feedback regarding how a new strategy might guide the future direction of genetic and genomic healthcare in Victoria over the next ten years.

## Vision

The health of all Victorians is optimised through clinically appropriate integration of genomics into health care.

## Mission

To integrate genomics into Victorian health care to better prevent, detect and manage disease and disease risk and predict and/or prevent the complications of disease and treatment.

## Objectives

1. To facilitate access for all Victorians to genomic knowledge to advance their health and wellbeing.
2. To contribute to Victorian health system sustainability through judicious use of genomic knowledge.
3. To better leverage existing research and development in Victoria.

## Principles

The September 2015 Victorian Health Reform Summit identified the following 10 key principles to guide future health reform:

* Person-centred care with equitable access
* Integration
* Prevention and early intervention
* Technology and data
* Workforce
* Transparency and accountability
* Evidence-based care
* Sustainability
* Innovation
* Medical research.

These key principles are encapsulated within the following proposed principles to guide genetic and genomic services:

1. Patients as partners.
2. Equity of access - equal response for equal need.
3. The health system needs to promote responsiveness and sustainability.
4. All decisions and actions need to be ethically defensible and acceptable to the community.

The mission, vision and objectives are illustrated in the following diagram:



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| ***Q2. Are the proposed Vision, Mission, Objectives and Principles appropriate to guide genetic and genomic healthcare developments into the future? If not, what should be changed?*** |

# Action areas

The following factors were identified earlier in this discussion paper as likely impacting the future provision of genetic and genomic healthcare in Victoria:

* Models of care and service system capacity
* Literacy;
* Personalised medicine including personalised prevention;
* Co-productive healthcare;
* Ethical, legal and social issues;
* Value for money.

The proposed priority areas for a genetic and genomic healthcare framework are:

* Engaging all stakeholders and building awareness;
* Clarifying the ethical and legal framework;
* Adopting the evidence;
* Building service system capacity; and
* Driving innovation.

For each of the priority areas, the following are outlined:

* Key issues;
* Steps already taken;
* Proposed actions over the next two years; and
* Issues for the longer term.

***Q3. Are the proposed priority areas the right ones to achieve the vision or are there other areas of higher priority that should be considered?***

# Engaging and building awareness

**Key issues**

* Good communication is central to both engaging stakeholders and building awareness of genetic and genomic health care.
* Genomic testing is being marketed directly to consumers and health professionals.

**Steps already taken**

The department’s internet site and the Better Health Channel have been updated to reflect current Government commitments and validated public information.

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| **Proposed actions** | **Expected outcomes** |
| 1. Work with consumer advocacy groups and other stakeholders to develop a robust communications and engagement strategy to raise public awareness of genomic health care and implement a genomics literacy program for the Victorian community.
 | A genomics literacy program for the Victorian community that is better informed and engaged to participate in genomic discussions. |
| 1. Scope a ‘Family History’ project to raise awareness of the genetic basis of disease and assist in developing personal and community responsibility for health and wellbeing.[[6]](#footnote-6)
 | Evidence on which to base a decision of whether or not to implement a family history project. |
| 1. Establish a trusted and accessible portal of information for the general public through the department’s Better Health Channel.
 | The general public use the Better Health channel as a trusted source of genetic and genomic information. |
| 1. Liaise with relevant clinical stakeholders, and their training bodies, to ensure that genetics and genomics are incorporated into undergraduate and post-graduate training and continuing professional development.
 | Improved awareness and understanding of genomics among health professionals. |

**Issues for the longer term**

The issue of sharing genomic information ‘in the public good’ is complex and requires broad and intelligent discussion with the community, scientific and clinical experts and policy makers ensure that a framework for Victoria reflects the values and desires of the community.

**Vignette 4**

A GP is asked by a patient to explain the results of a genetic test she had ordered from a US-based ‘direct-to-consumer’ genetic testing company. The results stated that the patient has an increased risk of developing Alzheimer disease. The patient is extremely anxious and expresses concerns about proceeding with her plans to have children because of the risk they will ‘inherit’ her risk.

***Q4. Are the proposed actions the right areas to achieve the vision or are there other actions of higher priority that should be considered?***

# Clarifying the ethical and legal framework

**Key issues**

* An individual’s genetic and genomic information has ramifications for his/her health and lifestyle choices, as well as those of relatives and future generations.
* A balance is needed between an individual‘s rights, community responsibility and health professionals’ obligations.
* Genomic sequencing raises issues around privacy for patients and family members.

**Steps already taken**

With Government support, the University of Melbourne Law School has contributed to consideration of the ethical, legal and social implications (ELSI) of clinical and research genomic and their implications for driving Victoria’s future economic advantage. This included assessing community expectations about governance of genomic information repositories with the Melbourne Genomics Health Alliance.

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| **Proposed actions** | **Expected outcomes** |
| 1. Support collaborative research of the ELSI of genomics in clinical practice.
 | Clear guidelines to manage personal genomic information**.** |
| 1. Work with stakeholders, including consumer advocacy groups, to develop a communications and engagement strategy to raise awareness about the ELSI of genomics.
 | Informed community discussion about the ELSI of genomics. |
| 1. Clarify the ELSI priorities for consideration and action both locally and nationally.
 | A list of priorities and relevant fora (local or national) identified to progress them. |
| 1. Advocate nationally for an update on the implementation, currency and scope of the Australian Law Reform Commission’s 2003 report *Essentially Yours: The Protection of Human Genetic Information in Australia*.
 | A national position on the adequacy of current protection for personal genomic information in Australia. |

**Issues for the longer term**

Policy responses to the ELSI of genomics must be grounded in clear, consistent and defensible principles.

Reactions to the ELSI of genomics must both respond to existing genomic technologies and their applications, and anticipate future discoveries and developments.

An active consideration of the ELSI should be integrated into genomic clinical, research and policy activities.

**Vignette 5**

A man in his early 20s was found to carry a genetic mutation related to Lynch syndrome, a condition associated with a higher risk of colorectal cancer. After applying for an increased level of life insurance coverage with three insurance companies, he was offered cover but with a cancer exclusion. In one application, the man informed the insurance company he considered his risk of colorectal cancer was the same as the rest of the population because he intended to undergo yearly colonoscopies to reduce his risk of developing precancerous polyps. After the man complained to the Australian Human Rights Commission, he was offered full insurance cover with a smaller exclusion clause. This illustrates potential implications of testing for, and finding, a genetic mutation, despite precautionary surveillance measures.[[7]](#footnote-7)

***Q5. Are the proposed actions the right areas to achieve the vision or are there other actions of higher priority that should be considered?***

# Adopting the evidence

**Key issue**

* New evidence should be quickly utilised to protect and improve the health and wellbeing of Victorians.

**Steps already taken**

The Victorian Government has committed $25 million over the next four years to pilot the use of whole exome sequencing in clinical practice through the Melbourne Genomics Health Alliance.

The Victorian Cancer Agency facilitates cancer research across Victoria, including study of germline and somatic cancer mutations.

The Victorian Government has developed strategies to support health and medical research, clinical trial access and growth in the biotechnology sector.

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| **Proposed actions** | **Expected outcomes** |
| 1. Evaluate the clinically and ethically appropriate role of genomic sequencing in clinical practice and infectious disease surveillance.
 | Provision of evidence to support the clinically and ethically appropriate adoption of genomic sequencing and knowledge into healthcare services. |
| 1. Develop a decision-making framework to support clinically and ethically appropriate integration of genomic knowledge into Victorian healthcare.
 | Safe and sustainable incorporation of genomic knowledge into healthcare practice. |

**Issues for the longer term**

International and national collaboration are essential if the full potential of genomic medicine is to be realised. This will require robust technology platforms and secure information sharing.

Victoria’s work will need to align with national policy work currently underway: the Commonwealth Government is leading development of a national genomics policy framework.

**Vignette 6**

A five month old infant presented with low muscle tone and poor feeding. Multiple blood and urine tests, lumbar puncture and an MRI brain scan indicated Leigh disease, a mitochondrial condition caused by mutations in many different genes, was a likely diagnosis, which causes a progressive decline in neurological function. There is no cure or treatment. The family was devastated to learn their baby’s condition would worsen with time and was unlikely to survive more than a few years. However, whole exome sequencing enabled a diagnosis of Thiamine Metabolism Dysfunction Syndrome 2, rather than Leigh disease. While this condition is progressive, treatment with thiamine and biotin may slow or prevent disease progression. The baby’s treatment was immediately altered and his parents were given the good news that his outlook is now quite positive. The baby’s older siblings were also tested as this condition can cause milder neurological deterioration in previously normal children. One sibling, found to have the same condition, immediately commenced thiamine and biotin to prevent any neurological deterioration.

***Q6. Are the proposed actions the right areas to achieve the vision or are there other actions of higher priority that should be considered?***

# Building service system capacity

**Key issues**

* Access to genetic and genomic services varies across Victoria.
* A flexible, multidisciplinary and adaptive service system is needed in the face of rapidly changing genomic knowledge.

**Steps already taken**

*Victoria’s Cancer Action Plan 2008-12* (VCAP) increased service system capacity and facilitated the establishment of permanent, on-site integrated cancer genetic services. The VCAP initiative was leveraged to further integrate non-cancer genetic services.

Paediatric and adult services for inborn errors of metabolism are now delivered by acute health services.

A statewide service stream design and infrastructure planning framework has been developed for the Victorian healthcare system in response to the Travis and Kings Fund reports. Work is being overseen by a Ministerial Advisory Committee.

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| **Proposed actions** | **Expected outcomes** |
| 1. Continue mainstreaming of genetic and genomic healthcare across the service system.
 | Genomics accepted as routine healthcare practice. |
| 1. Develop a statewide genetic and genomic services design and infrastructure service plan, including workforce capacity.
 | Clarity regarding future infrastructure and health workforce requirements. |
| 1. Develop a plan for the integration of pathogen genomics into the state-funded health system.
 | Priorities and resource requirements identified to facilitate clinically and ethically appropriate integration. |

**Issues for the longer term**

The health system will need to address the increasing awareness and utility of the role of genetics and genomics in improving health and wellbeing.

**Vignette 7**

A 45 year old woman was diagnosed with acute myeloid leukaemia (AML). Treatment options include a stem cell transplant, which requires the Human Leucocyte Antigen genes (HLA type) of potential donors and the recipient to be tested to identify immunological matching. If the HLA genes are not identical, there is a risk of lack of engraftment (i.e. rejection of the stem cells or graft versus host disease where the new stem cells react with the patients tissues). Both these complications can cause mortality. DNA sequencing of the HLA genes can identify minute genetic differences that ensure immunological matching of donor stem cells to the recipient and offer the best transplant outcome.

***Q7. Are the proposed actions the right areas to achieve the vision or are there other actions of higher priority that should be considered?***

# Driving quality, safety and innovation

**Key issues**

* Genomic healthcare needs to be safe and high quality.
* Victoria is established as a leader in genomic healthcare innovation.

**Steps already taken**

The 2016 *Review of Hospital Safety and Quality Assurance in Victoria* is expected to contain a number of recommendations to improve the quality and safety of health services in Victoria.

The Victorian Government recently launched its *Healthier lives, stronger economy: Victoria’s Health and Medical Research Strategy 2016-20, Victoria’s International Health Strategy 2016–2020* and *Future Industries Medical Technologies and Pharmaceuticals Sector Strategy*. The Victorian Government provided $25M to the Melbourne Genomics Health Alliance to build genomic sequencing capability in Victoria.

Further innovation in Victoria is being driven through the Better Care Victoria Innovation Fund, the Victorian Cancer Agency and clinical networks to test new ideas and develop additional capabilities.

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| **Proposed actions** | **Expected outcomes** |
| 1. Establish an independent advisory body to provide ongoing advice on Victorian genomic healthcare provision and drive adoption of innovative practice.
 | Key stakeholders advise on and drive service system change. |
| 1. Leverage existing state and national research, innovations and biotechnology funding to support translational research in genomic healthcare.
 | Victoria established as a national leader in, and major driver of, clinical genomic innovation. |
| 1. Position Victoria’s public health reference laboratories to take a national leadership position in the development and implementation of national microbial and infectious disease genomics policy and expertise.
 | Victoria established as a national leader in public health microbial and infectious disease genomics testing and practice. |

**Issues for the longer term**

There is an opportunity to position Victoria as a national leader in many aspects of genomic healthcare, including rapid translation to healthcare practice, pathogen genomics, resolution of ethical, legal and social implications and strong community engagement.

**Vignette 8**

A 47 year old woman with metastatic ovarian cancer was found to have a mutation in the BRCA1 gene. Clinical trials are open for a class of drugs known as PARP-inhibitors, which may be more effective in people with this gene mutation. These drugs are currently unavailable through the public health system in Australia but have been approved for some patients in the United States. Identification of this woman’s gene mutation allowed her to access to the drugs through a clinical trial with hope of a better outcome.

***Q8. Are the proposed actions the right areas to achieve the vision or are there other actions of higher priority that should be considered?***

# Next Steps

The Victorian Government is committed to pursuing excellence in care in partnership with patients, communities and service providers. Discussion points and vignettes have been provided to illustrate some of the issues and complexities in the field of genomic healthcare.

The Department of Health and Human Services seeks feedback on the content of, and questions in, this discussion paper. Your feedback will inform the development of a framework for genomic healthcare in Victoria. The framework is expected to be completed by late 2016. To ensure this happens, we request your feedback by **Friday 9 September 2016**.

You can provide feedback, using the template provided, in the following ways:

Mail: Genetics and Health Technology

 Department of Health and Human Services

 GPO Box 4057

 MELBOURNE VIC 3001

Email: genetics@dhs.vic.gov.au

Phone: (03) 9096 6126

Fax: (03) 9096 9115

Website: www2.health.vic.gov.au/hospitals-and-health-services/patient-care/speciality-diagnostics-therapeutics/genetic-services

1. ‘Genome’ refers to the entire complement of an organism’s genetic material. ‘Genome’ and ‘genomic’ are used in this sense throughout this Discussion Paper. [↑](#footnote-ref-1)
2. Muin Khoury, From Precision Medicine to Precision Public Health: challenges and opportunities, 26 October 2015, Annual Population Health Science Colloquium, Stanford Medicine, USA. [↑](#footnote-ref-2)
3. The workshop summary is available [here](https://www2.health.vic.gov.au/hospitals-and-health-services/patient-care/speciality-diagnostics-therapeutics/genetic-services). [↑](#footnote-ref-3)
4. Personalised medicine is the capacity to predict disease development and influence decisions about lifestyle choices or to tailor medical practice to an individual. National Health and Hospitals Reform Commission. *A healthier future for all Australians*, June 2009. [↑](#footnote-ref-4)
5. Co-productive healthcare – patients and professionals interact as participants within a healthcare system. Batalden et al. Coproduction of healthcare service, British Medical Journal Quality & Safety Online, 16 Sept 2015. [↑](#footnote-ref-5)
6. The USA uses a national public holiday when families congregate to encourage the collection of family medical history. [↑](#footnote-ref-6)
7. From: Keogh et al. *Medical Journal of Australia* 2013;199(5):363. [↑](#footnote-ref-7)