Response ID ANON-PR8U-2NNY-P

Submitted to **Draft National Preventive Health Strategy**
Submitted on **2021-04-18 19:52:35**

Introduction

1 What is your name?

Name:
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2 What is your email address?

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3 What is your organisation?

Organisation:
Australian Genomics

VISION

4 Do you agree with the vision of the Strategy?Please explain your selection. (1000 word limit)

Agree

Vision Text:

AIMS

5 Do you agree with the aims and their associated targets for the Strategy?Please explain your selection. (1000 word limit)

Agree

Aims Text:

The specific nature of the targets under each of the aims across the lifespan, equity and increased investment suggest they have been carefully considered using the breadth of data sources cited in the Strategy (such as AIHW, ABS National Health Survey).

PRINCIPLES

6 Do you agree with the principles?Please explain your selection. (1000 word limit)

Agree

Principles Text:

Multi-sector collaboration: The intention to enhance multi-sector collaboration to deliver this Strategy is well-supported by the boxes outlining related strategic guidance in each of the focus areas for action. Although about to undergo a renewal, the National Health Genomics Policy Framework (2018-2021) could also be considered in this context for areas of alignment.

Adapting to emerging threats and evidence: The breadth of emerging evidence for using genomic technologies in preventative health is summarised in response to question 9 of this consultation.

ENABLERS

7 Do you agree with the enablers? Please explain your selection. (1000 word limit)

Agree

Enablers Text:

Research and evaluation / monitoring and surveillance: Datasets need to be available for both government and academic-led research in a timely way. While privacy and data security cannot be compromised, an agile prevention system needs to be enabled by health data and health outcome monitoring. Here, the recent proposed changes to legislation led by the Office of the National Data Commissioner, and the recommendations of the Privacy Commission report on Data Availability and Use will be important. There is still a great deal of work to be done in this area, particularly in relation to harmonising datasets and their availability across jurisdictions. Australian Genomics initially applied for access to hospital inpatient and emergency datasets in each State and Territory of Australia to

provide a basis for economic evaluations of the value of genomic testing in different contexts. After 5 years of pursuing the data and consolidation to focus our efforts on collecting data from only three States, the application process is only now nearing completion. This experience is evidence of the need to address improvements in access to data.

Another key component will be investment in the development of a federated approach to linking health data, electronic medical and other health records, a wide range of test results (including genomic data) and other information such as environmental and socio-economic datasets. Harnessing these datasets and applying machine learning approaches would be needed for generating robust risk prediction scores. Having a federated approach to data sharing would be of immense benefit in facilitating preventative health strategies, particularly in our federated health system with established data systems and jurisdictional health privacy principles. The National Approach to Genomic Information Management Blueprint considers many of these issues and the principles may be adaptable to sharing other kinds of health data across jurisdictions.

8 Do you agree with the policy achievements for the enablers? (1000 word limit)

Agree

Enablers - Policy Achievements Text:

FOCUS AREAS

9 Do you agree with the seven focus areas? Please explain your selection. (1000 word limit)

Agree

Focus Areas Text:

Our earlier response to the consultation on the development of the NPHS provided evidence that the focus area “increasing cancer screening and prevention” may be too specific. There are many other diseases where screening and prevention can be done, and for which genomic technologies can be the foundation of those screening programs. This includes both rare and common diseases, including cardiovascular and other chronic illnesses, reproductive carrier screening and newborn screening. A broader focus area relating to health screening would allow a wider range of screening programs to be considered over the course of the 10-year Preventative Health Strategy.

Preventive screening for thousands of recessive diseases is the subject of the Mackenzie’s Mission: Australian Reproductive Genetic Carrier Screening Project. Mackenzie’s Mission is a $20M MRFF project aimed at determining the feasibility of providing free carrier screening to every Australian couple that wants it. The project will test thousands of couples for about 750 recessive and X-linked genetic conditions (conditions passed on to children from parents who are ‘carriers’ of the condition but who do not have it themselves). Outcomes including a proposed Medicare item number for expanded carrier screening are expected to lead to sustained implementation and will be an example of how the use of genomic medicine in preventative health can reach a significant proportion of the population. Genetic screening for recessive disease is a precision medicine preventive health measure, and couples can be helped to have healthy pregnancies following identification of increase chance of genetic disorders through the use of In Vitro Fertilisation (IVF) and Pre-implantation Genetic Diagnosis (PGD), medical technologies that are already in use in Australia. We are pleased to see that the strategy lists “reproductive/maternal” as one of the new focus areas for addition in Figure 7.

With such preventative screening measures, the need arises to expand genotype allele frequency in diverse groups. The goals of the recently established Centre for Population Genomics revolve around the generation and analysis of genomic data sets from thousands of Australians, and their linkage with clinical and biological information. Specific projects include the development of a new genome reference database, spanning diverse Australian communities, that improves our ability to interpret genetic changes found in Australians affected by genetic disease. NHMRC funding to the National Centre for Indigenous Genomics to develop Indigenous genomic representation has enrolled ~ 500 community members to date. Indigenous genomics will only progress with the involvement of the communities involved. This aligns with goals of the National Aboriginal and Torres Strait Islander Health Plan.

An emerging concept is that of “whole of life” genomics; starting with genomic testing in newborns, with serial reinterrogation of the genomic data throughout the life course as new medical questions arise. Newborn screening using whole genome sequencing could eventually replace the current heel prick tests, with the advantage of the genomic information then being available for emergency situations, to gain pharmacogenomic information (for example potential drug response), and for interrogation of other conditions relevant to a person’s stage-of-life.

Early and rapid diagnosis of genetic conditions in affected children using whole genome sequencing can change the course of management of the condition for the child, and in some cases can influence the prognosis significantly. In addition, finding out whether the disease-causing variant was inherited from the parents or occurred de novo, is powerful for informing future reproductive decisions for the parents (Stark et al., 2019). The Acute Care Genomics study was established in 2018 to implement and evaluate a comprehensive multi-centre network for ultra-rapid genomic diagnosis in the Australian healthcare system. In its first phase, the study provided ultra-rapid genomic testing to more than 200 critically ill babies and children from across 12 Australian hospitals, with results available to most families in just three days. In May 2020, the study team were granted $5M from the MRFF to build further capacity in all Australian states and territories. Investment in the capacity for early detection of genetic disorders will have major effects on the introduction of appropriate surveillance for predictable impacts on health as a consequence of the underlying disorder, on restoring reproductive confidence, and on eventual access to precision/targeted therapies. We are pleased to see that the strategy lists “infant/congenital” as one of the new focus areas for addition in Figure 7.

Application of polygenic risk scores (PRS) is still under evaluation and more evidence is needed to support the target populations who should receive risk reducing interventions, but Australia could learn from and build upon large-scale international studies already underway (such as the US NHGRI PRS Consortium and UK Biobank). In terms of informing screening programs, genomics has the potential to stratify people with common cancers for surveillance and treatment (e.g. breast, colorectal, prostate), as well as cardiovascular disease. Ideally, incorporating family history, environmental, genetic risk, epigenetic and other phenotypic factors into a stratified risk profile will mitigate any overdiagnosis and over screening. In this context, we are pleased to see that the strategy lists “cardiovascular” as one of the new focus areas for addition in Figure 7.

Having access to pharmacogenomic information would also have strong potential to improve health throughout a lifetime. Roughly 1.7 million Australians receive a medication each year for which, if available, pharmacogenomic information would be relevant. Widespread use of such information could significantly decrease emergency and hospital presentations related to adverse events, with up to 3% of all hospitalisations in Australia resulting from adverse drug events at an annual cost of $1.4B. There are up to 15 gene/drug interactions that already have evidence that would impact on prescribing to patients and would have significant health budget cost-savings. This could be implemented as another new focus area of the NPHS.

Actions around partnerships, research, evaluation and preparedness provide an opportunity for the preventative health strategy to align with other existing and future programs such as the 10-year, $5B MRFF Missions (Health Genomics, Brain Cancer, Million Minds Mental Health, Dementia, Ageing and Aged Care, Indigenous Health, Stem Cell Therapies, Cardiovascular, Traumatic Brain Injury).

10 Do you agree with the targets for the focus areas? (1000 word limit)

Agree

Focus Areas - Targets Text:

11 Do you agree with the policy achievements for the focus areas? (1000 word limit)

Agree

Focus Areas - Policy Achievements Text:

CONTINUING STRONG FOUNDATIONS

12 Do you agree with this section of the Strategy? Please explain your selection. (1000 word limit)

Agree

Continuing Strong Foundations Text:

As discussed in relation to consultation question 7, if academia is to be a prevention partner there needs to be a framework and mandate for better timely access to health data.

The inclusion of Figure 7 (the creation of new focus areas) provides good context for the direction of the Strategy, and Australian Genomics is pleased to see the representation in this figure in areas where genomic technologies and their emerging uses will be important, including cardiovascular, reproductive/maternal and infant/congenital.

FEEDBACK

13 Please provide any additional comments you have on the draft Strategy. (No word limit)

Comments Text:

We have summarised a strong case for the use of genomic technologies in our submission, and while not necessarily highlighted here, this extends to other ‘omics. The Strategy is weak in these areas, and the table briefly alluding to “genetic, epigenetics and telomere biology” in the Biomedical factors contributing to risk appears to cite general, outdated references.

Australian Genomics:
Australian Genomics is an Australian Government initiative supporting genomic research and its translation into clinical practice. Through broad engagement and a national collaborative approach, it achieves two key objectives: to improve efficiency, reach and timeliness of genomic research projects, and to support Commonwealth State and Territory health departments in the implementation of genomics research outcomes by refining and communicating evidence to inform policy development.

Australian Genomics engages with current and emerging government policy and priorities to identify gaps and opportunities, to support policy and action for integrating genomic technologies into the health system. By interfacing with consumers, governments, industry and global genomics initiatives, Australian Genomics drives change and growth in the sector.