Response ID ANON-PHPV-2MJ7-X

Submitted to **Stakeholder feedback on the draft WA Health Genomics Strategy 2021**
Submitted on **2021-01-31 08:20:15**

About you

In relation to the Strategy, which stakeholder group(s) best describes you?

Other (please specify below):

Other stakeholder group:
Submission by Australian Genomics, which represents a broad range of genomics stakeholders

Do you currently work in the WA public health system?

No

Which state or territory do you mostly reside in?

Not Answered

Other stakeholder location:
Australia-wide representation

Part 1: Draft Strategy

Do you have any comments or concerns about the vision?

Please provide further comments:
The vision seems somewhat time-limited, and there may be cause to consider whether the wording about “transformative potential” is still going to be appropriate toward the end of the 10-year Strategy? Evidence gathered in the first two horizons (up to 5 years) of the implementation plan will (hopefully) provide a great deal of evidence achieving the realisation of the transformative potential of genomics, at which point the transformation and translation phases will progress.

The vision and aim also do not touch on the 'appropriate' application of genomic technologies, in relation to their application where evidence supports utility and effectiveness, as well as improvements over standard of care. The term appropriate may also allude to when it is socially or culturally appropriate to use genomic technologies in healthcare.

It may be worth including both, or alternatively differentiating between, research and clinical aspects of genomics in the vision statement. As such, a reference to “clinical services” may be clearer.

A small but meaningful tweak to the vision to “maximise health genomics knowledge to appropriately transform clinical services to benefit all Western Australians” may be an alternative.

Part 1: Draft Strategy

Do you have any comments or concerns about the underlying principles? Are there any additional principles that you think should be included?

Please provide further comments:
Evidence gathering and research are significant themes of the Strategy, which run through the Priorities, Horizons and Actions of the Implementation Plan. Some of this evidence gathering and research may be incorporated into the principle of ‘Utility’, but not all genomic research is able to be measured by utility, particularly ‘basic’ research. It would therefore be nice to see a principle relating to research and/or innovation added to this list, to acknowledge its role in underpinning clinical genomics.

Whether ‘Equity’ and ‘Diversity and Inclusion’ need to be separate principles could be considered.

In terms of ‘Flexibility’, despite the rapid advancement of genomics and the need for strategic adaptation to progress efficiently, this also needs to be justifiable and appropriate – particularly in the controlled, accredited and standardised application of genomics in healthcare.

To asses whether these principles do underpin the planned implementation of the Strategy, a useful exercise may be to check off each of the principles as they align to actions items of the Horizon 1 Implementation Plan. This exercise may reveal specific principles that are underrepresented in the planned actions. In addition, some principles, if not acted upon from the beginning of the Implementation Plan, may be hard to "play catch-up” on, particularly those relating to trust, empowerment, diversity and inclusion.

Part 1: Draft Strategy

Do you have any comments or concerns about the enablers of success? Are there any additional enablers that you think should be included?

Please provide further comments:
Given the person and family-centric priority area, and equity, diversity and inclusion, empowerment and trust principles, consumer involvement could be added to the list of enablers. WA has a strong reputation for involving consumers in research, with Australia-wide consumer involvement strategy and policy influenced by the work of Anne Mackenzie and colleagues (Telethon Kids Institute). WA could capitalise on this wealth of local experience and knowledge to engage consumers in delivery of all aspects of the Implementation Plan.

A culture of research is an essential enabler for working toward shared goals through collaboration and innovation and is equally important in public sector and public/private partnerships. Other health strategies and frameworks have reported on how crucial a culture of research is, including for building research and clinical trials capability in public and private healthcare settings eg in the National Clinical Trials Governance Framework. Rather than a standalone enabler, this may come under the governance and senior leadership categories.

Commitment at all levels and by all stakeholders to the implementation of the strategy is another key enabler.

Reliable and ongoing resourcing is an important but not directly quoted enabler. This could be conflated with ‘senior leadership and support’, and is perhaps redundant as it is specified as Priority 2: Sustainable Investment.

Part 1: Draft Strategy

Do you think the key priority areas, goals, and initiatives are appropriate for guiding the scope of work to be undertaken to achieve the Strategy’s vision? If not, please explain below.Are there any additional priority areas, goals, or initiatives that you think should be included?

Please provide further comments:
These priority areas are common with most other genomics-related strategic policy frameworks. This is a logical way to align with and leverage of work that is being undertaken by other states and nationally, given the clear alignment of these priorities with the National Health Genomics Policy Framework (NHGPF). The WA Strategy building upon the priority areas of the NHGPF with the addition of priorities 5 and 6 may signal areas where the NHGPF could be built upon during its revision in 2021.

With the aspirational goals of the WA Digital Health Strategy 2020-2030, incorporating those goals in this Strategy could solidify WA as a leader in digital health in genomics. Central to this will be the delivery of telehealth services for genomics, particularly for rural, regional and remote communities. Availability and linkage of patient data will be necessary for the evidence gathering research that is a priority of the first two Horizons of the Strategy.

WA is leading research and innovation in many areas of genomic medicine, so it is good to see as a priority area.

One concern is that the Strategy itemises ‘ initiatives’ under each priority area, but these do not align with the implementation plan’s ‘actions’ in the survey information and questions that follow.

A suggested reordering of the priorities in a more logical, sequential order:
3: Services
1: Person- and Family-centredness
2: Sustainable Investment
4: Workforce
5: Digital Health and Data
6: Research and Innovation

Part 2: Implementation Plan

Horizon 1 (first 2 years): Developing the blueprint and early priorities

Do you think the focus of Horizon 1 is appropriate for the implementation of the Strategy? If not, please explain below.

Please provide further comments:
Developing a blueprint for genomics and the evidence gathering described in Horizon 1 will need to be underpinned by cooperation, shared goals, data availability and research. This is where building a culture of research (a suggested enabler) could drive action toward a common goal of developing a full picture of the current ‘state-of-play’.

To achieve this in first few years will be difficult, given the notoriously slow processes for research ethics / governance and data availability and access for research. This will not change rapidly, so WA should be actively exploring what evidence being generated by other states can be applied to WA. One example is the extensive health economic, implementation science and workforce and education evidence coming out of the last five years of the Australian Genomics research program. For example, the comprehensive health economic body of work that demonstrates the value of genomic testing across traditional methods (cost-effectiveness and clinical utility) as well as reaching into areas including willingness to pay and family impacts of genetic disorders. A list of these research outputs is available and this would be highly relevant to the evidence gathering phase of the Strategy’s implementation.

Australian Genomics’ work through 18 different clinical ‘flagship studies’ or disease areas has demonstrated that for some cancer types, the results of genomic sequencing can inform future care in up to 85% cases. Rapid genomic testing in the acute care setting for critically ill infants has found a genetic diagnosis in around 50% cases – one of the highest and most consistent diagnostic rates for genomic sequencing reported internationally. This evidence could be applied to the research and service planning phases of the Strategy in the WA context.

Planning for increasing workforce, up-skilling the workforce and service capacity should start in Horizon 1 rather than Horizon 2, especially where new educational streams are to be established. WA may be able to use existing national plans to fast–track planning for these areas. For example, a national mapping project has already been conducted on the genetic workforce – this could be updated for WA personnel, rather than repeated (doi: 10.1002/jgc4.1101).

Rather than a 'point in time' evaluation, establishing a continual monitoring/evaluation framework to measure the impact of the implementation / changes real time will be important to measure success of the strategy, and make changes as needed.

The audit / evaluation should also conduct a landscape analysis of what is available nationally that can be adapted / repurposed for WA. There is so much activity in other jurisdictions / national initiatives in these priority areas WA should leverage the investments of others and strive for harmonisation of approaches.

Horizon 2 (2-5 years): Building momentum

Do you think the focus of Horizon 2 is appropriate for the implementation of the Strategy? If not, please explain below.

Please provide further comments:
Evidence building about the benefits genomics can deliver seems to be a focus of the first two Horizons (5 years) of the 10 year plan. Whether this represents an over-emphasis on evidence gathering phases should be considered, since the benefits of genomics are being reported with rapidly increasing frequency (see also answer to last question), and much of the data could be applicable to the WA context.

Horizon 3 (5+ years): Realising the transformative potential of genomics

Do you think the focus of Horizon 3 is appropriate for the implementation of the Strategy? If not, please explain below.

Please provide further comments:
It is interesting that the question preamble suggests that predicting how genomics will progress over time is difficult. We recently read with interest the "bold predictions" for the advancement of human genomics by 2030 put forward by Green et al., (doi.org/10.1038/s41586-020-2817-4). Some of those predictions may be unachievable in the public health system within that time, or even not desirable, but it is likely that in Australia we will see:

- Price of genome sequencing and the cost of data storage will come down to the point where it may be more cost-effective to re-sequence than to store data for long periods.
- WGS will replace standard newborn screening tests.
- Reproductive carrier screening will be accessible to all those who want it.
- Pharmacogenomic information will incorporated into a person's health record, available in My Health Record or its future equivalent.
- Cancer tumour profiling will be available to all people affected by cancer, to inform diagnosis, prognosis and treatment.
- Higher rates of genetic diagnosis for rare diseases will be achieved.
- Polygenic risk scores will stratify the population for risk of developing common diseases and inform those for whom surveillance strategies will be usefully applied.
- Infectious disease genomics and Public Health genomics will be priority areas.

Looking to the MRFF GHFM Roadmap and international genomic strategies like that of NHS would inform the areas where genomics will progress over the next 10 years. Those areas should be identified and planning on how to incorporate those into WA Health should start now. Indigenous people and other priority groups will not be able to share in these benefits if the important engagement work and reference genome representations are not achieved. WA has a significant role to play in this given its Indigenous population and remote population.

The commercialisation of genomics is a complicated area which should be pursued only in a way that builds and maintains public trust.

Part 2: Implementation Plan

Do you think the actions for this priority area are appropriate for focussing efforts and resources in the first two years? If not, please explain below.Are there any additional actions (relevant to this priority area in the first two years) that you think should be included?

Please provide further comments:
Action 1.4: The adoption of the Australian Clinical Genomic Testing Consent Form should be done with careful consideration, as it is not necessarily consistent with facilitating the data availability that will be required to achieve the goals of Priority 6 (research and innovation). This is because the Consent Form strategy is to seek consent to share genomic information for research purposes separately (and potentially on a separate occasion to the clinical consent process). The risk is that the person will then never being asked for, or give, consent to research participation, due to the increased administrative burden that would be involved, or lack of ongoing contact with the family. This in turn limits discovery research and will ultimately impact diagnostic rates if information about normal variation and pathogenicity of genetic variants is not shared. The Global Alliance for Genomics and Health (GA4GH) supports the imperative nature of responsible genomic data sharing, and genomic data sharing for research aligns with the FAIR data principles. Recently, Dame Sue Hill, the CSO of the NHS spoke about the infinity loop between genomic research and clinical genomics in her plenary address at the Festival of Genomics and Biodata (January, 2021). The Australian Genomics National Clinical Consent Form shares many elements with the Australian Clinical Genomic Testing Consent Form, but includes research participation as a separate tick box consent on the clinical test consent form. This amount of separation between research and clinical consent is likely to satisfy the NPAAC guidelines for obtaining consent to research use of genomic data.

Action 1.5: In developing a digital hub, WA should look to consumer and clinician education materials that have recently been developed. For example, information for health consumers could be repurposed from the materials developed by Australian Genomics at genomicsinfo.org, while information for professionals about genomics has been developed by Melbourne Genomics (learn-genomics.org.au). The Centre for Genetics Education and many international organisations have a developed high quality information for consumers. Liaising with the Centre for Population Genomics, a joint Garvan/MCRI initiative, would be mutually beneficial in relation to developing consumer information for priority populations.

It was surprising to find no mention of rural and remote communities in the implementation plan and that Aboriginal people are only mentioned in relation to developing a digital hub (action 1.5). The strategy document mentions the need to champion equity of access to 'priority populations' however this isn't really translated into the Implementation Plan. From our Australian Genomics study in 2019 on areas of unmet need in the access to clinical genomic testing, we know these are populations of concern in relation to unmet need and require active consideration. The Implementation Plan would benefit from an action point relating to assessing unmet need and the subsequent development of strategies to address them with the stated intention to evaluate and amend interventions as required.

Part 2: Implementation Plan

Do you think the actions for this priority area are appropriate for focussing efforts and resources in the first two years? If not, please explain below.Are there any additional actions (relevant to this priority area in the first two years) that you think should be included?

Please provide further comments:
The background information to Priority 2 (on page 11 of the Strategy document), regarding the issues and challenges being faced in WA, was very informative and set the context well – however, perhaps would be better placed in the accompanying Discussion Paper rather than in the Strategy?

Action 2.1: describes the collection and 'awareness of' data on benefits? Perhaps it would be useful to expand on whether this is referring to awareness by policy and decision makers at health system level. Also in relation to this action is the concern about research ethics / governance and data availability and access, and how this will impact achieving such an action within a two year timeframe. Drawing upon previously published papers would contribute greatly to locally acquired evidence (eg Tan et al., JAMA Pediatr, 2017, 17, 855-862; Catchpool et al 2019, Genet Med, 21, 2815-2822). Australian Genomics can also provide a list of published outputs on the health and economic benefits of genomic medicine, from the last five years.

Action 2.3: The test directory is an excellent action and would be a resource that could be leveraged and built upon nationally.

Part 2: Implementation Plan

Do you think the actions for this priority area are appropriate for focussing efforts and resources in the first two years? If not, please explain below.Are there any additional actions (relevant to this priority area in the first two years) that you think should be included?

Please provide further comments:
Action 3.1: WA could compare and collaborate with other state-wide services in Australia to develop a state wide plan. A good example would be Genetic Health Queensland, which, like WA services a large geographical area and remote populations, Indigenous people, and successfully incorporates Telehealth services.

Action 3.2: Measures of performance and quality included could include demand, and an evaluation of the appropriateness of test ordering and diagnostic rates, to determine whether they are in line with current national and international standards. This will be increasingly important as non-genetics specialists order more genomic tests. Additional measures could include result turnaround times, and patient experience of genomic testing.

Action 3.3: Meeting demand is a concern that could be at least partially addressed by innovative approaches to support genomics services, including developing decision aids, online education and consent. An example of this would be investigating whether an adaptation of the Mackenzie's Mission Australian Reproductive Carrier Screening project recruitment process would be appropriate for future reproductive carrier screening services. Again, looking to the other states' pathology services as possible models for WA, including the sequencing services offered by Pathology Queensland.

Action 3.4: In addition to the evaluation, reporting and ongoing assessment framework identified in this action, a role for implementation science methods to also be applied here should be explored in shaping the detail of the Implementation Plan.

Action 3.5: There is already evidence to suggest that clinical genomic testing is not being offered equitably. For example, a survey of clinical genetics and genomics services led by Australian Genomics in 2019 revealed that one of the main challenges was that specific populations are unable to access genomic testing, or are not presenting to genetics services. These populations included Indigenous people, non-metropolitan populations and those with intellectual disability. Australian Genomics is building on this work with a follow up study that will audit Australian laboratories offering genomic tests (panels, exomes, genomes) and compare testing with population distribution accessing those services over a 6-month period, via a postcode audit.

The State of the Nation Blood Cancer in Australia report published by the Leukaemia Foundation comprehensively highlighted the many areas of inequity and the metro/regional divide in terms of access to services, diagnosis and treatment, as well as psychosocial impacts of cancer. Included was the reported figure that regional people are 7% less likely to receive a genetic or genomic test to guide diagnosis, and 7% less likely to receive a genomic test to guide treatment, than metropolitan based survey participants. These equity issues need to be addressed before the ability to a share in the benefits of genomics widens any further between different groups in Australia.

Other more general observation in relation to this priority area include the opportunity for WA to build a business model that can capitalise on an international reputation for excellence in diagnosis of NMD, and as a developing hub for the delivery of reproductive genetic carrier screening. WA also has strong potential to be a leader in the incorporation of RNASeq and other 'omics in the personalised medicine toolkit.

Planning investments in a service for the future should look to and align with the research outcomes that will be emerging from GHFM funded research - with the draft Roadmap and Implementation Plan now published, and several funded projects already underway, WA should start to consider how those findings will be translated into the WA health service context.

Furthermore, emerging areas of clinical genomics service delivery should be built into budgets upfront, including for costs for routine sequence re-analysis, individualised return of results and incorporation of personalised decision-making through digital consent platforms.

Part 2: Implementation Plan

Do you think the actions for this priority area are appropriate for focussing efforts and resources in the first two years? If not, please explain below.Are there any additional actions (relevant to this priority area in the first two years) that you think should be included?

Please provide further comments:
The Australian Genomics Workforce and Education program has conducted numerous research projects over the past five years, mapping the genomic workforce and education landscape and investigating current practice plus education and training needs across different professions and specialties.

In particular, our national surveys of genetic specialists – clinical geneticists and genetic counsellors – and other medical specialists not trained in genetics include responses from Western Australian health professionals that may provide useful insights for the evidence-gathering in Horizon 1. These data could inform Priority 4 (workforce) as well as Priorities 3.1 (delivery models) and 6.1 (clinical, diagnostic and research collaborations). The Australian Genomics Workforce and Education program is willing to share WA-specific data to support the Implementation Plan of the Genomics Strategy.

In addition, we convene the Genomics Education Network of Australasia, a community of practice with over 120 members across Australia, which may be relevant to Priority 4.2 (knowledge sharing).

Action 4.1: The workforce challenges were well outlined and discussed in the Strategy and Discussion Paper. As well as training more professionals, other solutions should be considered. For example, is there a way to alleviate data entry from Genetic Counsellor role? Can digital tools support the genetic counselling process, as well as test ordering or test reporting process? Decision aids are showing great potential for use by patients considering genomic testing, as well as to assist in clinical decision-making. Can additional support staff be recruited to help prepare for MDT meetings? Can patient liaison officers take on some of the patient facing roles? Many of these suggestions could also be considered in the context of the Digital Health and Data priority.

Action 4.2: There are many opportunities to avoid redundancy and duplication of other efforts that are mature in Australia in developing genomics knowledge. WA non-genetics specialists should participate in test ordering credentialing being developed by HGSA.

Other actions should include building upon the Indigenous health workforce; considering scholarships/fellowships with a “return home” component on completion of study to attract qualified professionals back to WA, and WA should also engage with other states including NSW, where strategies are already being developed to increase the workforce in genomics related fields.

Part 2: Implementation Plan

Do you think the actions for this priority area are appropriate for focussing efforts and resources in the first two years? If not, please explain below.Are there any additional actions (relevant to this priority area in the first two years) that you think should be included?

Please provide further comments:
Action 5.1: For guidance on sharing, storage and protection of data, WA should look to GA4GH for data standards and governance endorsed by the international genomics community. Australian Genomics has incorporated GA4GH standards and Australian guidelines, policy and principles into the governance processes for its cohort genomic and health data, and all policies are publicly available on the Australian Genomics website. This ensures data is stored securely, access is provided for ethically approved uses, and data is standardised to ensure future interoperability and usability.

Actions 5.2 and 5.5: In relation to cloud storage and a data governance frameworks, in 2020 Australian Genomics surveyed domestic (Australian) genomic data infrastructure managers and users, as well as representatives of international genomic initiatives, to find out about the current landscape of genomic data infrastructure, and to learn about the requirements and preferences for data storage in the future. The report is available to stakeholders involved in the survey, including PathWest. Australian Genomics is willing to share the survey templates to be adapted to the WA context to facilitate the collection similar information, which we recommend due to the highly valuable information and collective perspectives we received from stakeholders. Data storage frameworks will be informed by the National Approach to Genomic Information Management, developed under the PRG. Developing a cloud infrastructure needs national involvement: formation of a national panel of genomic data experts to evaluate and align activity, as well as use collective bargaining when engaging with cloud providers.

WA should also review and consider adopting the many digital and data tools developed by Australian Genomics. WA is already participating in Shariant, a platforms for sharing clinically curated variants among laboratories nationally. Other tools that could be adapted to clinical genomics services include those to standardise phenotype capture (which relates to Priority 5.4), pedigree tools, electronic test ordering and CTRL, a digital dynamic consent platform.

Part 2: Implementation Plan

Do you think the actions for this priority area are appropriate for focussing efforts and resources in the first two years? If not, please explain below.Are there any additional actions (relevant to this priority area in the first two years) that you think should be included?

Please provide further comments:
An efficient research ethics and governance system will be key to achieving any actions relating to this priority.

The Strategy and Discussion Paper seemingly focus on translational research. However, ‘basic’ research is equally important and warrants adequate and continued funding. There are many examples where technique development, functional genomics research, gene discovery research and ELSI research all underpin translational genomics research. Another priority research area should be that which improves equity – building Aboriginal genome representations, and investigation into genetic conditions in Indigenous people eg diabetes risk.

Research priority setting in the first two years should be done with consumer and other stakeholder participation, and should aim to capitalise on the research priorities of the GHFM and international initiatives. For example, pharmacogenomics is an emerging as a priority internationally, as the health and health economic benefits are clearly demonstrated.

Action 6.1: Seeks to promote collaborative genomics related research focused on the consumer and health system priorities (which is to be commended) however there is no mention of implementation research or health economics which will be essential to facilitate overcoming barriers, learning lessons from implementation and shaping future implementation along with generating the economic argument in line with consumer views. There would be value in broadening the research focus outlined in the implementation plan to include implementation and health economic research. “Knowledge of the critical factors of success, key gaps and current capacity and capabilities” as stated in Horizon 1 will only be possible with implementation and HE research.

Action 6.3: Availability of genomic and health related data will be for research will be diminished if the Australian Clinical Consent Form is implemented, as discussed in relation to Action 1.4.

Other feedback

Do you have any further comments on the draft Strategy?

Please provide further comments:
Overall, the documentation is very good. The Discussion Paper, particularly, is candid and transparent, which bodes well for the finalisation and implementation of this strategy. However, our overall concern is that the document describes a strategy that suggests WA will pursue the body of work to be developed in the Implementation Plan independently. Any evaluation / landscape analysis needs to incorporate efforts in other jurisdictions / from national initiatives, both to leverage the experience and investment of others, and to align with national and global positions. Australian Genomics would be pleased to have the opportunity to work with the WA Strategy team to identify opportunities to leverage Australian Genomics outputs and to highlight the progress of other individual and organisational partners in our network.

The documentation refers to the PRG and the NHGPF in the current tense – to our understanding the PRG will not be reconvened, and activity associated with the NHGPF implementation has either been completed up or abandoned.

Follow up... almost done!

Would you like to ...

Be contacted about your feedback if we would like additional information or clarification?, Receive a copy of the final Strategy?, Be contacted about opportunities to provide feedback on the implementation of the Strategy?

If you ticked any of the boxes above, please provide your contact details below.

Please provide further comments:
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