

## Feedback: University of Sydney

### Draft NSW Health Genomics Strategy –Feedback Form

Comments are welcomed on the draft NSW Health Genomics Strategy. Please provide feedback by completing the relevant sections below and return to Ms Alex Scott, Senior Policy Officer, Specialty Service and Technology Evaluation Unit, Health System Planning and Investment Branch, NSW Ministry of Health at [alexandra.scott@moh.health.nsw.gov.au](mailto:alexandra.scott@moh.health.nsw.gov.au) by **COB Tuesday 25 April 2017**.

#### Section 3 – The Promise of Genomics

Does this section provide sufficient context for clinical genomics?

Please detail any additional content you would like to be considered.

This section gives useful context for clinical genomics, and indicates current actions at the policy level which is a relevant starting point.

#### Section 4 – Genomics in NSW: Part of the plan

Does this section provide appropriate examples of foundational drivers for genomics in NSW?

Please provide details of other **existing** examples you are aware of.

There are other **major existing foundational drivers based at the Sydney Children’s Hospitals Network that are servicing very large clinical needs in Clinical Genomics in NSW** and they should be included:

##### Section 4.1 Improving health through clinical innovation

- Add to paragraph 2 (Genetic rare diseases):

The Faculty of Medicine, University of Sydney is highly connected with all of the LHDs where there are Clinical Schools of the Faculty. The University connections have facilitated the infrastructure for many research programs that have contributed to the current leadership of several NSW groups in genomics. This includes the Sydney Genome Diagnostics Laboratory, Western Sydney Genetics Program, The Children’s Hospital at Westmead (CHW), Sydney Children’s Hospital Network (SCHN) Health District. This laboratory, headed by Associate Professor Bruce Bennetts, is providing **NATA-accredited genomic testing** for patients with genetic rare diseases of the kidneys, eyes, bones, and immune and neurological systems. This laboratory offers testing for patients from throughout NSW and nationally, and also receives international requests. This work has resulted in many examples of new clinical diagnoses and new genetic information for families, leading to changes in clinical management (e.g., Ref 1). This laboratory is currently the largest provider for molecular tests for rare diseases in NSW.

Other laboratories, not already mentioned, which are associated with the University of Sydney through Clinical School and LHD connections include the Molecular Genetics Laboratory at Royal Prince Alfred Hospital (RPAH) led by Professor Ron Trent, and the Neurogenetics Laboratory led by Professor Garth Nicolson at Concord Hospital.

#### **Section 4.1 Improving health through clinical innovation**

- Add to paragraph 4 (Models of care):

The Faculty of Medicine, University of Sydney, is highly connected with all of the LHDs where there are Clinical Schools of the Faculty. Hence, the University connections facilitate the infrastructure for many clinical research programs and teaching that have contributed to the current leadership of several NSW groups in genomics. This includes the contribution from the Discipline of Genetic Medicine which is a cross-cutting Discipline of the Faculty of Medicine.

Clinical Genetics Services at The Children's Hospital at Westmead (CHW) conducts regular multidisciplinary clinics attended by Clinical Geneticists, Genetic Counsellors and disease specialty experts. Clinical Genetics Services at other Clinical Schools and LHDs including those of the Northern and Central Sydney Clinical Schools and Nepean and Westmead Clinical Schools, also conduct assessment and delivery of complex genomic results to a wide variety of patients. Almost all of these staff have a University of Sydney affiliation including library membership which crucially facilitates rapid and free access to the latest literature in genomics, which is not currently provided to staff by NSW Health. At these clinics, genomic results from the Sydney Genomic Diagnostic Laboratory, CHW, and other national and international laboratories are used for clinical care for patients. Complex genomic results that feed to these clinics for implementation of patient care are reviewed and discussed at fortnightly multidisciplinary Genome Variant Analysis Review (GVAR) meetings at SCHN.

Multidisciplinary clinics at CHW include as follows, and all of these clinics also receive referrals from all parts of NSW and interstate:

Connective Tissue Diseases Clinics (CHW) - attended by Endocrinologists, Rehabilitation Paediatricians, Clinical Geneticists and Genetic Counsellors;

Genetic Eye Clinic (CHW) - attended by Ophthalmologists, Orthoptists, Clinical Geneticists and Genetic Counsellors, and networked with other Ophthalmological services at Sydney Eye Hospital and Westmead Hospital;

Neurogenetics Clinic (CHW) – attended by Neurologists, Physiotherapists, Clinical Geneticists and Genetic Counsellors

Genetic Metabolic Diseases Service (CHW and SCH) – attended by Metabolic Paediatricians with Clinical Genetics training, Metabolic Nurses, Genetic Counsellors.

The Clinical Genetics Services at CHW also provides service to Adult Genetic Clinics at Westmead Hospital. The Clinical Genetics Services at all of the LHDs associated with Clinical Schools of the University of Sydney also provide service to Prenatal Clinics at Westmead Hospital, RPAH, RNSH, and Nepean Hospital. At these clinics, genomic test results are interpreted and used for patient care.

#### **Section 4.2 Ground-breaking research**

These exemplar leading research programs have arisen due to close interactions between Medical Research Institutes, Universities and LHDs. The examples of leading genomic research and technological capabilities in NSW may also include Gene Therapy research capability at the Children's Medical Research Institute in liver disease and gene therapy vector development, research in Neuromuscular disease at the Kid's Research Institute and Cardiac research at the Centenary Institute. The value of the interactions between the MRIs, Universities and LHDs is critical and recognition and enhancement of these relationships would be beneficial in promoting genomic health care in several ways in NSW, as has been recognized by NHMRC. As an example, in 2015, the NHMRC recognised Sydney Health Partners (SHP) as one of four Advanced Health Research and Translation Centres (AHRTCs) across Australia. Information about this partnership should be added to Section 4.5, Collaborations and Partnerships.

#### **Section 4.5 Collaborations and partnerships**

Sydney Health Partners is an additional significant collaboration in NSW, with genomics as a cross-cutting theme, for facilitation of genomic medicine in NSW.

In 2015, the NHMRC recognised Sydney Health Partners (SHP) as one of four Advanced Health Research and Translation Centres (AHRTCs) across Australia. It comprises several local health districts, including Sydney Children's Hospitals Network, Northern Sydney, Sydney and Western Sydney, the University of Sydney and its Clinical Schools in these areas, and related research institutes including Children's Medical Research Institute, Kolling Institute, Centenary Institute, Anzac Research Institute, and Westmead Institute of Medical Research. Genomics is one of the cross-cutting themes facilitating communication across these different institutions between clinical and laboratory leaders, and researchers, academics and undergraduate teachers working in the genomics area. This represents an established network of clinical, research and teaching expertise in genomics, and future strategy should build on these connections.

One of the genomics projects of SHP is the development of an ethical and governance framework to facilitate genomic, functional genomic and therapy-based research for rare diseases across LHDs, Universities and MRIs by developing an umbrella "Program of Research" in genetic rare diseases. This Program of Research is now approved, with the Specific Project application process for disease-groupings underway. This work is necessitating development of paths through the ethical and governance frameworks across several institutions in NSW. The experience gained from this work will be valuable in working towards an overarching framework to better facilitate clinical and research genomic medicine.

## Section 5 – The vision for NSW Health genomics

Does this section adequately describe the collective vision for genomics in NSW?

Are there any other factors for consideration?

The vision for NSW Health genomics is well-described.

Two minor suggestions for consistency with the foundations for the vision developed in Chapter 4.

- In the graphic at the beginning of section 5.1 Realising the Vision (p.18) the text box bottom left of the “Strengthen the Foundations” text box should read “World class research”. The word research should be restored, as this is critical to underpin many elements of realization of the vision.
- Also, for consistency, the heading at 4.2 “Ground Breaking Research” (p.12) should be “World Class Research”.

## Section 6 – Delivering the vision

Do the recommendations capture the scope of work that needs to be undertaken in NSW?

Are there any additional recommendations for consideration?

Are there any further challenges that should be identified?

Please provide your comments to these questions in the relevant sections below.

### 6.1 Leadership and Governance

Provision of leadership at the state level for clinical genomics in NSW is absolutely critical for execution of the vision.

This governance body needs to include representation from contributors in the various content and geographical areas. A key performance indicator for the leadership and governance arrangements ought be the efficacy of access by researchers to genomic technology, and the support that researchers receive in using this technology.

In recommendation 1, it is unclear what is meant by “appointment of a subject matter expert co-chair”. Does this mean someone with expertise covering many of the relevant areas, or are subject matter experts to be co-opted as co-chair for particular areas?

**As an additional comment, the “Delivery of the Vision” section does not appear to encompass capacity for research in genomic medicine.**

While delivery of what is currently known in genomics would be an excellent achievement, it is important to recognize that there is still a requirement for research in many areas to maximize genomic medicine health outcomes in the short, medium and long term. This includes further research to uncover additional genotype-phenotype correlations, and disease genes and their functions for the patients and families where these are not currently known. It also includes research in genetic therapies which will have the capacity to delivery new treatments for patients with genetic disease where none were previously available. Other research areas related to genomic medicine include health economic, legal, ethical and educational aspects.

The University of Sydney, often in collaboration with NSW Health, has traditionally been at the forefront in contributing to many research endeavours in genomics and other areas of medicine. We would recommend that Section 6 also include reference to research components in each of the points related to delivery of the vision, or alternatively there could be a separate point about research in genomic medicine in this section. The University of Sydney has significant expertise in many of these areas, including the Faculties of Medicine and other Health Sciences, and the Faculties of Science and Law.

## 6.2 Focused application: clinical need, validity and utility

Enhancement of focused application on the basis of clinical need, validity and utility would be an extremely welcome step forward in genomic medicine in NSW. This would provide an immediate and significant clinical benefit. It would also set the groundwork for more widespread advances in clinical application.

There is also a need for ongoing research in concert with this step, to evaluate the clinical validity of the genomic testing, and also, for example, to apply the new technology to various disease phenotypes to determine detection rates where these are not currently known, and discover novel disease genes and variants. A strong collaborative relationship with the Universities, including the University of Sydney, will facilitate ongoing leadership in the best and most appropriate focused applications of genomics.

## 6.3 Service delivery: commissioning and utilising genomic technology

The issues raised here indicate the large scope of work required for the best possible provision of genomic medicine in NSW.

**Integration of genomic medicine into mainstream care** is an important goal that needs careful consideration and resourcing.

For this to be delivered, there will need to be increased resourcing to enable genetics health professionals contribute to relevant specialty and primary clinics, for assistance with accurate delivery of genomic care.

Simultaneously, to achieve this goal, there is a strong need for increased education in clinical genomics of the current and future healthcare workforce. This will need significant enhancement of current teaching in clinical genomics at the undergraduate, postgraduate and professional levels. This will need enhancement of funding for teaching in genomics at the Universities and through the LHDs and professional bodies. A strong collaborative relationship with the Universities, including the University of Sydney, will facilitate ongoing leadership in the best approaches and implementation of teaching in clinical genomics across several areas of the genomics workforce.

The University of Sydney's current strategic plan includes a program whereby the institution is establishing a number of core research facilities. These research facilities have a campus wide remit to engage with researchers at Sydney and beyond, and via NCRIS and other linkages, offer a portal to national and international initiatives. Sydney Mass Spectrometry, and Sydney Informatics Hub are two of the University's core research facilities that could play a part in the implementation of a service delivery model.

#### 6.4 Genomic data and infrastructure: handling 'big data'

All of the issues indicated here are relevant.

An additional point that needs to be added is the need for increased education in genomics for the healthcare workforce to be able to use and appropriately interpret the "big data" delivered to them. This will also require increased deployment of genetic health professionals to assist disease-specific specialists and primary care physicians in interpretation of the "big data" for appropriate and relevant genomic care.

The University of Sydney, has leading expertise and investment in the area of "big data" analysis. A strong collaborative relationship in this area, will facilitate ongoing leadership in the best approaches and implementation of teaching and research in "big data" related to clinical genomics across several areas of the genomics workforce.

#### 6.5 Preparing the workforce for genomics

This section addresses the significant needs for increase in the genomics healthcare workforce, as well as the educational requirements for all healthcare providers.

One of the current major deficiencies is staff available for teaching of clinical genomics to undergraduates, postgraduate and professionals in healthcare. This teaching traditionally largely relies on genetic health professionals in the LHDs. Since clinical genetics itself is a relatively new and small specialty which only mainly operates in the public system (compared with for example, other more traditional specialties such as cardiology), there are only a limited number of genomic healthcare professionals available for teaching. In addition, with the current explosion in genomic medical care, these professionals are under significant time constraints due to the very large and increasing patient-care loads under which they are operating.

NSW Health is committed to supporting increased genomic literacy (Section 6.5), so a major useful initiative would be increased collaboration with the Faculty of Medicine, University of Sydney, which already has the infrastructure in place across several Clinical Schools and LHDs for approaches in genomic teaching. This could be increased across all of NSW in collaboration with other Universities and LHDs providing teaching in this area. A useful strategy would be to provide additional clinical academic positions in clinical genomics which would have the dual benefit of increased availability of clinical service for integration of genomics to additional multidisciplinary clinics, and increased availability for teaching and research to reach all members of the healthcare workforce.

## 6.6 Community engagement: maintaining public trust and confidence

These factors are relevant for achievement of the vision.

## Section 7 – Moving forward

What are your thoughts on the proposed way forward for the Strategy?

The proposed way forward will be a significant advance in implementation of clinical genomics in NSW.

An additional major benefit would be to include capacity for research considerations and recommendations across each of the five sub-committee pillars. So, in addition to providing guidance and advice on policy, regulation, ethics and investment, another horizontal plank could be “Provision of advice regarding strategy in research for future improved genomic healthcare”. This would additionally propel NSW Health with its University and Industry partners as a major visionary in genomic medicine.

## Sections 8- 10 – Glossary, Appendices, References

Please provide any comment or additions for inclusion.





Thank you for taking the time to comment on the draft NSW Health Genomics Strategy. Your feedback is valuable and will inform the development of the final Strategy document.