Planning for the Future

NSW Health Pathology Genomics Strategic Plan 2016-2018

NSW GOVERNMENT Health Pathology

Contents

1. Executive Summary

As a direct result of genomic technology, the health system is entering a new era of personalised medicine, based on accurate diagnosis and focussed therapy. This is being referred to as the genomics revolution.

With its current expertise in genomics, NSW Health Pathology (NSWHP) has a unique opportunity to position itself as NSW's premier provider of high quality, clinically relevant and professional genomic diagnostic services to health providers and their patients. In achieving this, it can become a leading contributor to national and international innovation in the area of genomics and personalised medicine.

The Genomics Strategic Plan 2016-18 sets out the goals, aspirations and initiatives for NSWHP Genomics over the next 3 years. This strategic plan aligns with both the NSWHP Strategic Plan 2014-18 and the current NSWHP Operational Plan.

The Genomics Strategic Plan 2016-18 will position NSWHP to:

- More effectively marshal its expertise and resources that exist across the organisation
- Reduce unnecessary duplication
- Create a strong unified and recognisable force in the marketplace
- Embrace new genomic knowledge and technologies as they relate to patient care
- Provide a broader range of genome testing based on high clinical utility to reduce the need for expensive outsourcing
- Attract external referrals for genome testing to support NSWHP's service delivery
- Position NSW Health Pathology as a leader in delivering state-of-the-art genomic diagnostics

The major initiative to be implemented to achieve these aims is the establishment of a statewide genomics service. The NSWHP Executive Leadership Team (ELT) has endorsed a preliminary structure for this service, which has been included as Appendix 1.

2. The Current Environment

2.1. Definition of Genomics

The term Genomics is commonly used to describe the study of an organism's total genetic complement or genome, including all of its genes and intervening DNA sequences.

For the purpose of this plan it also extends to the use of technologies commonly employed in the study of genomes to situations other than the study of a complete human genome. This incorporates cancer genomes, infectious organism genomes and partial genomes with targeted assays.

Medical Genomics is the application of genomic technologies to the care and management of patients within the hospital and community environments.

2.2. Overview of Genomics Landscape

In Australia, both the public and private pathology sectors provide diagnostic genomic testing, with the majority of complex genomic testing performed by the public sector. This situation is likely to change rapidly over the next few years due to technology changes, reductions in costs and wider clinical application of genomics. The two leading private pathology providers, Sonic and Primary Health, have established medical genomics programs.

Large research institutes, universities and private companies also provide selected genomic testing for specific applications. In NSW the Garvan Institute of Medical Research has partly used philanthropic donations to build capability for human whole genome sequencing.

The availability of public genome testing varies across the states and is primarily based on an individual laboratory model rather than a statewide model of service delivery. Exceptions are South Australia and Western Australia which operate statewide Pathology services (SA Pathology and Path West respectively) with significant genomics components.

Funding for genome related testing in NSW comes from Medicare rebates for a small number of lower complexity tests such as those for fragile X, haemochromatosis, Von Hippel-Lindau gene test and cytogenetic abnormalities identified by karyotyping and more recently microarray analysis. However, the majority of complex genomic testing does not attract a Medicare rebate and relies on either state funding, such as discipline specific clinic funding, or charges to private patients.

Direct to consumer genome testing is also increasing with samples being sent to overseas laboratories for analysis. Current demand for this testing is largely unquantified.

Nationally, the Australian Health Ministers Advisory Council (AHMAC) through its Hospitals Principal Committee (HPC) established the Genomics in Clinical Practice Working Group (GCPWG) to consider issues related to the broad introduction of clinical genomics in Australia. This group is considering policy needs in areas such as education, training and genomic literacy; pathways to ensure fair access to clinical genomic services; infrastructure and computing resources; and workforce development.

Internationally, there are several major national initiatives for genomics, most notably Genomics England, created under the UK Department of Health, and the Precision Medicine Initiative in the United States. These initiatives reflect the future importance and promise of genomics in medicine and help articulate the case to be engaged proactively in this area of pathology.

2.3. Current NSWHP Service Profile

Within NSWHP, four networks and approximately 10 laboratories provide around 87,000 genomic related tests per year. Sixteen percent of these tests are referred out to interstate, international or other private providers within Australia.

Genomic testing ranges from single target genotyping through to exome sequencing. NSW Health Pathology does not at this time have significant human whole genome sequencing capacity however; it is inevitable that it will provide such testing in the near future either through collaborative efforts or its own resourcing.

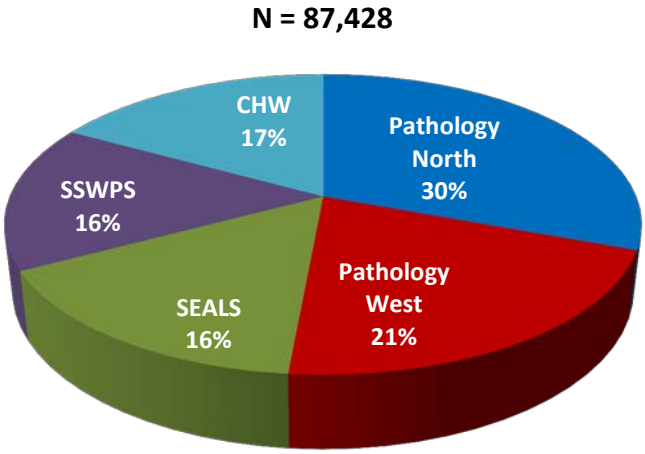
Testing can be grouped into five broad categories of oncology; rare diseases; cytogenetics; haematological non-malignant disorders; and pharmacogenomics. The complexity of genomic tests varies widely with some tests requiring considerably more resourcing to perform and report compared to others.

The majority of resources are currently directed to cytogenetic, rare disorders and oncology testing. Infectious diseases / public health genomics is at pre-clinical implementation at this time. However, there is a strong case for improving the accuracy and speed of drug resistance detection in difficult to culture pathogens that cause life-threatening bacterial, viral or fungal infections such as Mycobacterium tuberculosis. NSW Health Pathology laboratories have been implementing genome sequencing for drug-resistance testing, infection control and public health laboratory surveillance.

Testing by Network

Overall test referrals by Network are similar with the exception of Pathology North, which contributes 30% to overall test numbers. Figure 1 presents the overall test numbers for the NSWHP and Children's Hospital at Westmead (CHW) Genomics laboratories.

Figure 1: Total Genomics Testing Activity (2014/15)



Historically, the testing distribution relates to demand from Local Health Districts and resident expertise. While a significant degree of specialisation has occurred, duplication does exist with multiple services providing similar testing. This is particularly the case for less complex testing and some complex testing such as breast cancer genetic screening. Large panel Massively Parallel Sequencing (MPS) for rare diseases are also duplicated. Haematological oncology testing is performed in most networks and even at multiple laboratories within the same network.

Testing by Category

- Rare Diseases testing is primarily conducted by SEALS and CHW.
- Location of cancer testing is dependent on the type of testing provided. Testing for inherited cancer predispositions is primarily performed at Pathology North Hunter and testing for somatic variation in tumours is performed at several sites including SSWPS, Pathology North and SEALS.
- Haematological oncology testing is performed predominantly at Pathology North, SSWPS and Pathology West. However all networks contribute significantly.
- Non-cancer haematological testing such as that for Haemochromatosis is distributed across all networks
- Infectious disease testing including public health and hospital outbreak genomic testing will be implemented within the next 1-2 years and pilot studies are currently underway in both areas. Pathology West has commenced in-house sequencing of Listeria and salmonella for the detection of community outbreaks. The network has participated in the national initiative to harmonise whole genome sequencing of pathogens with epidemic potential through the Public Health Laboratory Network of Australia.

3. Drivers for Change

The NHMRC Strategic Plan 2013 – 2015 lists 'Preparing Australia for the 'omics' revolution in health care' as one of the major health issues to be addressed during the triennium:

> "The rapid advances in genomics and in a wide range of other 'omics' have already begun to emerge from the laboratory to directly affect patient care. 'Omics' is a shorthand way of referring to the steps in translation from the gene sequence through intracellular steps, into health and ill health of individuals. The potential for better focussed individual treatments and preventive strategies, and the implications for health policy and practice, are immense."

An expert opinion paper prepared in November 2015 by the Genomics in Clinical Practice Working Group (GCPWG) entitled, "Implementing clinical genomics in Australia" outlines the future for clinical genomics:

> "Genomic technology is revolutionising healthcare delivery and ushering in an era of precision medicine based on accurate diagnosis and focussed therapy. Continued development of genomic technology has driven improved accuracy and rapidly declining costs compared with previous genetic testing technology. More broadly, genomics is recognised as a disruptive technology that will reshape clinical practice. In the absence of consensus best practice guidelines and government policy, jurisdictions around Australia are using genomic technology in a clinical setting to varying degrees. This is creating unprecedented need for coordinated strategies to implement genomics into routine clinical practice, and ensure equity of access to clinical genomics for all communities across Australia".

In assessing the readiness of the Australian health system for the opportunity presented by genomics, the working group notes

> "(the assessment) highlights a health system that is unprepared for clinical genomics, with inequitable consumer access, inadequate workforce and training programs, a range of different funding mechanisms, inadequate and fragmented infrastructure and resources, and, a lack of comprehensive governance and regulation."

The NSWHP Genomics Strategic Plan identifies key priorities to address these issues and to ensure NSWHP keeps pace with the genomics revolution.

The plan outlines the initiatives that will address these priorities in the areas of service configuration, funding, workforce, informatics, technology and equipment, and partnership.

3.1. Service Configuration

Service Configuration Challenges

- The current model of service delivery is fragmented with little coordination of activities across the four networks or between laboratories within the same network.
- The Network centric model for service delivery does not allow coordination of testing or appropriate resourcing for the delivery of a state-of-the-art genomics service across NSW Health Pathology.
- Competition from the private sector requires us to focus on efficiency and productivity of our services to ensure we are providing contestable value for money services.
- Genomics covers a diverse range of tests and any reconfiguration of services and how they are delivered must consider the pressing timeframes and limited access to samples that exist for some genomic tests, for example somatic cancer testing.
- Local relationships with individual clinicians, (multi-disciplinary team meetings) MDTs, other pathology disciplines, other hospital clinics, and clinical trials units are critical.
- The relationship between clinical genetics and other medical disciplines varies from hospital to hospital.
- New applications are continually being identified for Genomics and often require extensive development and validation prior to implementation.
- Changing technology and clinical practice means that a reference or specialised test of today may be standard care tomorrow.
- There is an increasing availability of “direct to consumer” genomic testing with no, or minimal, medical gatekeeper role.
- Competition from other organisations is increasing, including research institutes who are introducing service delivery models for genomics that provide funding for research.

Strategic Implications

- Discussions about reconfiguring Genomics services will consider statewide models of service delivery, new technologies and changing demands for Genomic tests without discounting investment in, commitment to, and range of Genomic testing currently performed.
- This will require us to consider options for standardisation and consolidation of services and tests, and the development and implementation of new tests.
- It will be important to define the value proposition of public pathology genomics in a contestable market to highlight the unique strengths NSWHP can offer.

3.2. Funding

Funding Challenges

- There is no single financial view of genomics across NSWHP, making it difficult to identify opportunities for efficiency gains.
- The absence of appropriate funding models for reimbursement leads to inequitable access to genomics testing.
- NSW Health Pathology has a heavy reliance on external funding for equipment and testing.

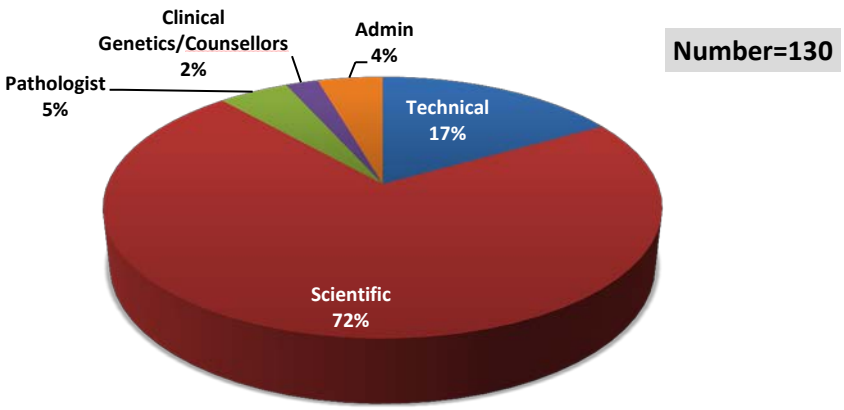
Strategic Implications

- A coordinated, statewide financial model will increase opportunity for efficiency gains and enhance accountability.
- Robust costing techniques for genomics, including how tests are to be reimbursed, will be essential to financial sustainability.
- Creating a sustainable and fundable Capital Asset Plan for genomics will support service planning and research activities.

3.3. Workforce

NSW Health Pathology employs approximately 130 staff in Genomics related testing and the CHW employs 61 staff. The CHW service, in addition to cytogenetics and molecular genetics, offers a newborn screening program and a biochemical genetics program. Figure 2 provides a breakdown of staff classifications across NSWHP (excluding CHW staff).

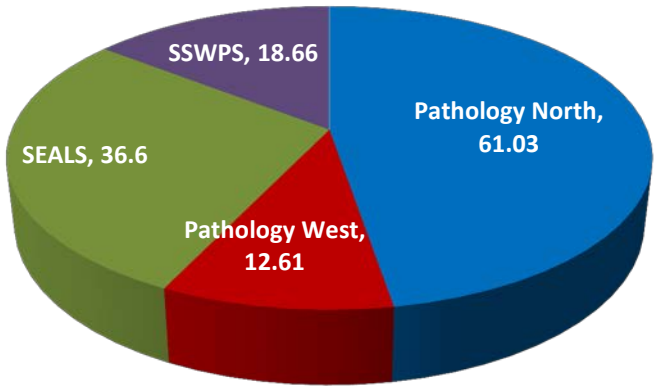
Figure 2: Staff Numbers and Classifications for NSWHP Staff (2014/15)



Pathologists receive training through registrar and advanced trainee programs in genetics, anatomical pathology, haematology, microbiology, chemical pathology, immunopathology and forensic pathology. The majority of pathologist training programs (other than genetics) contain little genomics content. There is a growing demand for pathologists with genomics expertise to participate in molecular MDTs.

Scientific and technical staff receive their training through universities and technical colleges. They are encouraged to obtain on the job training in Genomics, through either the HGSA (Human Genetics Society of Australasia) or the RCPA (Royal College of Pathologists Australia). Professional recognition for Clinical Scientist is gaining momentum through the RCPA. Figure 3 presents the location of staff working in NSWHP Genomics (excluding CHW staff).

Figure 3: NSWHP staff working in genomics by location (2014/15)



Workforce Challenges

- With relatively few technical officers / assistants and administrative positions, senior scientific staff are performing a range of scientific and non-scientific duties. This is taking scientists away from delivering the genomics service.
- There are limited non-managerial senior scientist positions for the development of Genomics and its applications to healthcare.
- Significant skill gaps exist, particularly around informatics. Currently there are no dedicated bioinformaticians in the NSWHP genomics workforce.
- There are no funded rotational training positions for pathologists or scientists to complement the small number of funded Genetic Pathologist positions.

- There is a need to train discipline specific specialists in genomic medicine relevant to their specialties such as anatomical pathologists, haematologists, microbiologists, chemical pathologists and immunologists to enable participation in molecular MDTs.
- There is an urgent need to promote training in both cytogenetics and molecular genetics to produce genomic scientists.

Strategic Implications

- The development of a genomics workforce will facilitate and support the merging of disciplines such as cytogenetics and molecular genetics into medical genomics and provide expert input into other pathology disciplines including anatomical pathology, haematology and microbiology.

3.4. Informatics

The term informatics is used in this plan to describe the control of test processes, analysis of raw data from instruments and the interpretation of complex data essential to creating a genomics report.

The large number of variants identified by large panel, exome and genome sequencing need to be annotated, curated and stored in clinical standard databases to produce reports and contribute to the knowledgebase.

NSWHP laboratories currently use a combination of Pathology LIS (Laboratory Information Systems) not specifically designed for genomics for test process control, multiple third party software solutions for analysis and annotation of data, and excel spreadsheets or generic databases for storage of data.

Informatics Challenges

- General pathology LIS do not meet the functional requirements for a genomics LIS or provide the functions required for genomic variant curation and clinical standard databasing of genomic data.
- Local Health District IT systems (infrastructure and firewalls) and NSW privacy restrictions inhibit cloud based storage or computing.
- Data sharing is a critical component for a statewide genomics service to leverage existing expertise.
- The current situation does not meet existing demands in many areas and is not sustainable for future genomics service delivery.
- The use of different third party software at multiple sites contributes significantly to expenditure and inhibits sharing of data.

Strategic Implications

- Statewide genomics informatics encompassing both laboratory information systems and bioinformatics will enable data sharing across NSWHP Genomics and promote national and international collaboration to produce best practice solutions.
- Informatics systems specifically designed for Genomics are urgently required for the efficient and safe delivery of Genomics services in NSWHP.
- Cloud-based solutions are highly desirable and will require detailed risk management assessments prior to implementation.

3.5. Technology and Equipment

Individual laboratories and networks have independently acquired technology and equipment for genomics testing. This has resulted in significant discrepancies between, and even within, services.

Additionally, many services rely heavily on funding sources external to NSW Health Pathology to provide genomics assets. Examples of this are outsourcing of high-performance computing and large panel MPS. Figures 4 presents an analysis of genomics assets for 2015/16. Figure 5 presents an analysis of number and age by network.

Figure 4: Analysis of NSWHP Genomic Assets (2015/16)

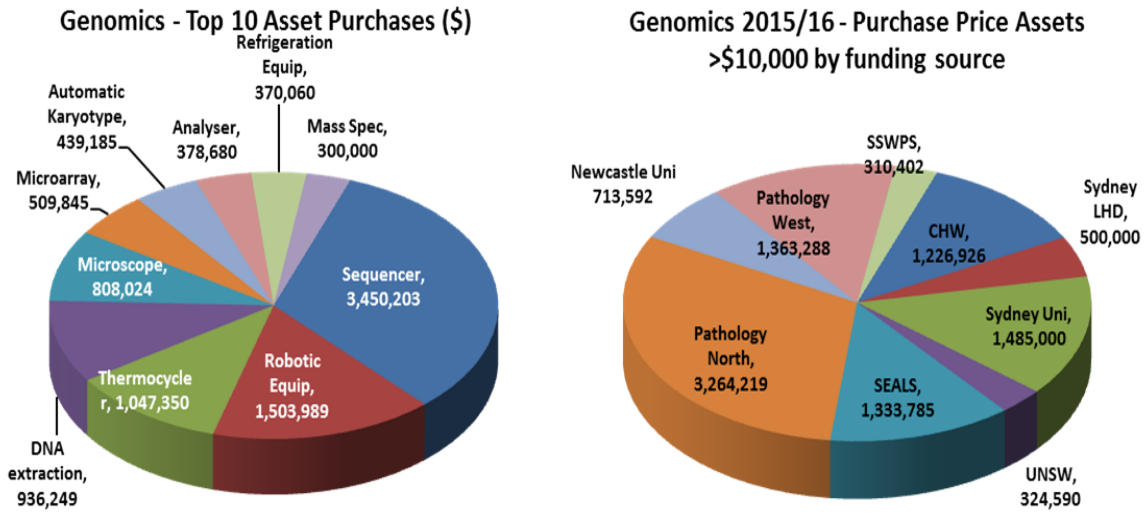
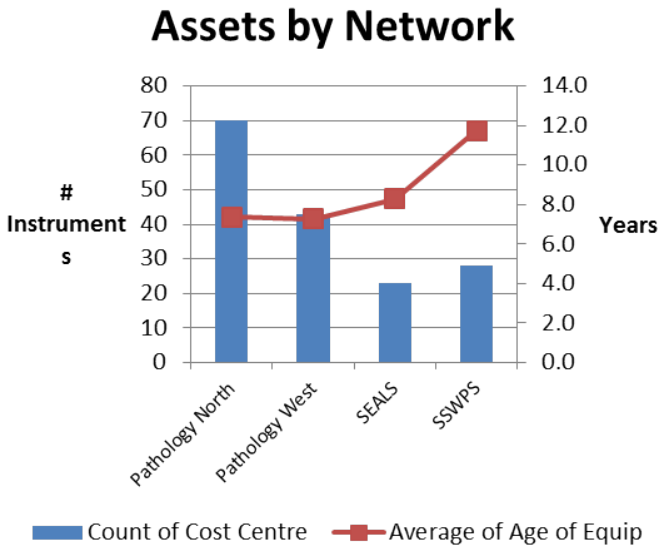


Figure 5: NSWHP Assets by Network



Technology and Equipment Challenges

- NSW Health Pathology has a heavy reliance on external funding for equipment and testing.
- There is no statewide asset management program for genomics.
- A disconnect exists between service delivery plans and resource / asset allocation.
- NSW Health Pathology does not have a mechanism to assess and respond rapidly to new disruptive technologies and processes in genomics.
- There is no whole of life cost approach to assessing future investment.

Strategic Implications

- Ongoing management of both resources and assets will be essential to maintain a strong technological position in Genomics through cost effective maintenance, timely replacement plans and investment in new technologies when the opportunity presents itself.
- A genomics asset strategy will support genomics service reconfigurations and provide a consistent approach to investment and management across NSWHP.

3.6. Partnership

In a fast moving and new technology discipline such as genomics, the most effective way to deliver a genomics service is through comprehensive partnerships with major referral clinics, key research institutes, State and Commonwealth governments, and relevant national and international genomics initiatives.

Partnership Challenges

- Translational research and increasing test complexity in genomics require a strengthening of existing clinical connections and new models for collaboration between pathology and clinical services.
- There are an increasing number of medical disciplines with a stake in genomics beyond traditional clinical genetics.
- Current delivery of genomics related testing relies heavily on existing connections with Local Health Districts, specialised referral clinics and research collaborations.

Strategic Implications

- Strong relationships with major referral clinics, key research institutes, State and Commonwealth governments, and relevant national and international genomics initiatives will support faster responses to changing technology in genomics.
- Strong relationships will also ensure strong clinical connections are maintained with both LHD and external referrers to NSWHP and foster formal collaborations with relevant research initiatives and national genomics programs.

4. The Future

4.1. NSW Health Pathology Strategy Map

Our Vision: Leading through innovation and collaboration to deliver excellence in service and outcomes

Our Purpose: Creating better health and justice systems

Strategic Directions

Patients, clinicians, LHDs and external customers

- Appropriate and timely access to services in rural, regional and metropolitan areas
- Quality, patient-centred, customer-focused services
- Innovative models of service delivery and practice

Partnerships, networking and processes

- Developing and maintaining strong clinical, scientific and research links
- Designing and participating in education, teaching and training opportunities to support and enhance services
- Working constructively with all key stakeholders
- Delivering consistent, integrated and transparent business processes and systems
- Driving continuous quality improvement

Resource accountability

- Managing and improving our performance
- Delivering contestable value for money services
- Managing our resources well
- Planning for the future

Our people, culture and capability

- Attracting, recruiting, developing and retaining quality, competent staff
- Leading research and translating outcomes into practice
- Effectively communicating, consulting and collaborating
- Creating a positive culture aligned to our values and purpose
- Ensuring a safe working environment

4.2. NSW Health Pathology Genomics Strategy

The NSW Health Pathology Genomics Strategic Plan 2016-18 cascades down from the NSWHP Strategic Plan.

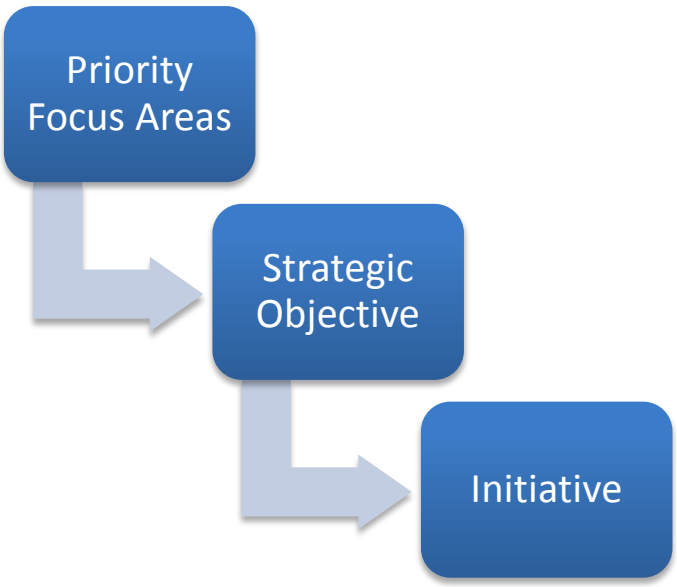
The NSWHP vision is ‘leading through innovation and collaboration to deliver excellence in service and outcomes’.

NSWHP Genomics embraces this vision and has responded to it with the following statement:

NSWHP Genomics is NSW’s premier provider of high quality, clinically relevant and professional genomic diagnostic services to health providers and their patients. It is a leading contributor to national and international innovation in the area of genomics and personalised medicine.

The tables on the following pages outline the NSWHP Genomics strategic objectives in each priority focus area (perspective). These strategic objectives mirror the strategic objectives of the NSWHP Strategic Plan.

Each strategic objective includes detailed initiatives that support the delivery of the NSWHP Genomics Strategic Plan 2016-18.



Patients, clinicians, LHDs and external customers: Customer Perspective
C1: Appropriate and timely access to services in rural, regional and metropolitan areas
C1.1: Deliver equitable access to appropriate and effective diagnostic genomics services, with particular focus on rural and regional areas, and vulnerable / disadvantaged populations.
C2: Quality, patient-centred, customer-focused services
C2.1: Implement a statewide service model and streamline the decision making processes across NSWHP Genomics. C2.2: Develop consolidated and coordinated genomic service nodes based upon current strengths and expertise. C2.3: Implement a single, consistent Quality Assurance and clinical governance framework for NSWHP Genomics.
C3: Innovative models of service delivery and practice
C3.1: Develop a preferred model of care for the delivery of genomics services: o Embrace new technologies and knowledge in genomics to deliver enhanced services that are highly integrated with the patient care process. o Develop new models for the diagnostic / clinical interface and handover of information, which are appropriate for complex genomic testing.

Resource accountability: Financial Perspective
F1: Managing and improving our performance
F1.1: Implement standard systems for the recording of tests through consultation with NSWHP Performance Support Team. F1.2: Establish systems for monitoring the performance of a statewide genomics service.
F2: Developing contestable value for money services
F2.1: Implement a coordinated, statewide financial model for NSWHP Genomics. F2.2: Implement robust costing techniques for genomics testing.
F3: Managing our resources well
F3.1: Establish an agreed policy for the investment / disinvestment in specific genomic tests in clinical practice. F3.2: Ensure appropriate delegations are in place to streamline NSWHP Genomics operations.
F4: Planning for the future
F4.1: Ensure that the statewide service model supports the development of genomics as a key enabler in molecular pathology. F4.2: Develop a NATA accredited service stream targeting research (including universities, research institutes and private organisations) and research infrastructure (including the NSWHP biobank and other biobanks). F4.3: Contribute directly to broader efforts to recognise genomic testing in the MBS. F4.4: Develop new national and global markets for the expertise of NSWHP Genomics. F4.5: Create a sustainable and fundable Capital Asset Plan for NSWHP Genomics.

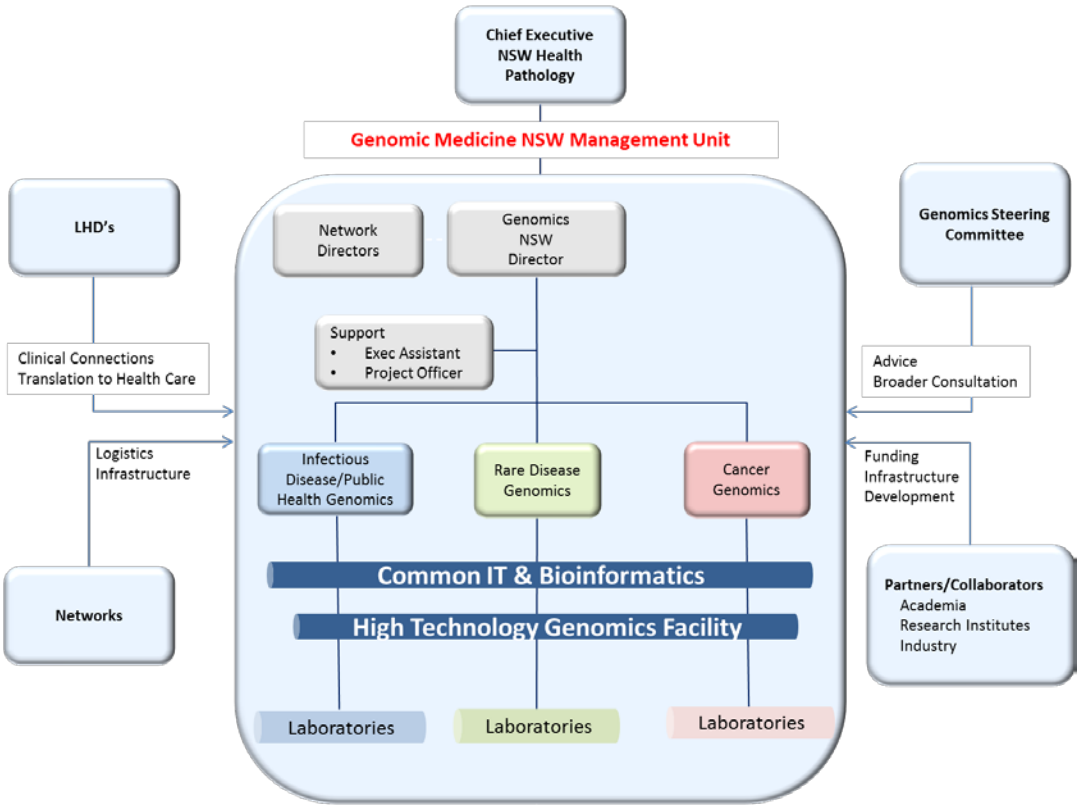
Partnerships, networking and processes: Internal Process Perspective
IP1: Developing and maintaining strong clinical, scientific and research links
IP1.1: Develop and prioritise plans for the application and integration of genomic technology into other NSWHP disciplines including anatomical pathology, microbiology, haematology, chemical pathology, immunopathology and forensic pathology. IP1.2: Facilitate and systemise clinical connections between Ministry of Health (including ACI, CINSW and others), LHDs and NSWHP Genomics to ensure they are: o Strong o Replicable / scalable o Sustainable IP1.3: Develop a Collaboration Plan for NSWHP Genomics. IP1.4: Participate in NSW, national and international genomics initiatives (including the NSW Genomics Reference Group and the Australian Genomics Health Alliance) in order to strengthen the position of pathology services and provide health leadership for genomics.
IP2: Designing and participating in education, teaching and training opportunities to support and enhance services
IP2.1: Develop an education and training strategy for NSWHP Genomics. IP2.2: Strengthen capacity of NSWHP Genomics workforce in areas of emerging technology and genomic techniques.
IP3: Working constructively with all key stakeholders
IP3.1: Develop a stakeholder engagement plan for NSWHP Genomics that integrates engagement, communication and change management activities. IP3.2: Strengthen integration and support of genomics activities undertaken by Children's Hospital at Westmead Pathology with Genomics NSWHP.
IP4: Delivering consistent, integrated and transparent business processes and systems
IP4.1: Develop contemporary, legally compliant and cost effective IT systems to support the needs of genomics (data collection, analysis, interpretation, bioinformatics, storage and dissemination).

Our people, culture and capability: Employee Learning and Growth Perspective
ELG1: Attracting, recruiting, developing and retaining quality, competent staff
ELG1.1: Develop a workforce strategy for NSWHP Genomics. ELG1.2: Develop the capacity of NSWHP Genomics and NSWHP in bioinformatics.
ELG2: Leading research and translating outcomes into practice
ELG2.1: Develop a research and innovation plan for NSWHP Genomics, which links to the NSWHP Research and Innovation Framework. ELG2.2: Define the commercialisation and industry partnership pathways for NSWHP Genomics, with reference to the broader framework for NSWHP.
ELG3: Effectively communicating, consulting and collaborating
ELG3.1: Develop an external marketing and communication plan for NSWHP Genomics. ELG3.2: Develop an internal communication plan for NSWHP Genomics.
ELG4: Creating a positive culture aligned to our values and purpose
ELG4.1: Develop strategies to support workforce equity and diversity among genomics employees.

Appendix 1: Preliminary Structure

The NSWHP ELT has endorsed a preliminary structure for statewide service delivery and financial models.

The proposed structure involves the creation of a Genomics Management Unit to manage the transition from the current model to a statewide service model and streamline the decision-making processes across NSWHP Genomics:



The three service nodes consolidate our five broad categories of current testing and add to these the emerging area of pathogen genomics. The current financial structure consists of 13 cost centres located in 4 networks. The financial reporting and performance management of the cost centres is the responsibility of the networks.

In this approach, genomics will be a unique entity and cost centre structure within the NSWHP reporting framework. Where appropriate, budgets will be transferred from the networks to genomics services following agreement by all stakeholders. Actual expense and revenue items will need to be transitioned within the same financial periods.

A single genomics entity provides centralised financial and workforce performance reporting across the state. A coordinated statewide financial model for NSWHP Genomics will provide much greater opportunity for efficiency gains, revenue realisation and improved resource utilisation and asset management.