Making good on the promise of genomics in healthcare: the NSW Health perspective

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ABSTRACT

NSW Health is implementing genomics as a mainstream component of clinical care. The strategic, holistic approach is considering infrastructure, data governance and management, workforce, education, service planning and delivery. This work is generating insights about how to realise the promise of genomics in healthcare, highlighting the need for strong foundations, real-world application, accessibility and a focus on people using genomic information in clinical care.

Keywords: clinical pathway, education, framework, genetics, genomics, implementation, infrastructure, NSW, omics, pathology, policy, precision medicine, translation, workforce.

Medical genomics is the application of genome-based knowledge to benefit human health.\textsuperscript{1} Such knowledge is the foundation of precision medicine, which uses an individual’s genomic makeup alongside environmental and lifestyle information to deliver targeted disease prevention, screening, diagnosis, prognosis or treatment.\textsuperscript{2,3} Precision medicine may help avoid unnecessary treatments, with benefits for patients, families, the health system and economy.\textsuperscript{4,5}

Recent decades have seen significant advances in genomic sequencing technology and a growing understanding of the interplay between genetic, environmental and lifestyle factors in disease, health and wellbeing.\textsuperscript{6} Cheaper, faster sequencing technologies improve availability and affordability of genomic tests. However, realising the value of genomics requires high-quality clinical interpretation and appropriate, acceptable application of data generated for the net benefit of patients and families.

NSW Health is committed to embedding clinical genomics into healthcare, using genomic information for disease surveillance, diagnostics and treatment (Fig. 1). The NSW Genomics Strategy\textsuperscript{4} is supported by implementation plans,\textsuperscript{7,8} a translation framework\textsuperscript{1} and education resources.\textsuperscript{9} Implementation, supported by state and national funding, involves collaboration between clinicians, policymakers, researchers, consumers and industry. This activity is generating valuable insights about how to realise the promise of genomics in healthcare.
To embed clinical genomics in mainstream clinical care NSW Health is prioritising three key areas for action.

1. Incorporating genomics into clinical pathways

Integrating genomics into mainstream clinical practice requires equitable access to tests and timely sharing of information and data generated for clinical and research use. With multiple organisations and stakeholders developing and funding genomic technologies, the potential for niche tests with restricted availability is significant. The NSW Genomics Strategy is developing realistic service delivery models to suit the NSW public health system and ensure genomic testing is available in regional, rural and remote areas. Examples include simplified ordering and consent systems, and cloud-based repositories to store and share data. Implementation activity is underpinned by knowledge of the NSW health system, patient journeys and service delivery models. Pilot projects in neurofibromatosis and genetic testing for intellectual disability have generated evidence about how to embed genomics safely and equitably in clinical care. Diagnosis of rare diseases is most beneficial if linked with clinically actionable outcomes and available treatments. This includes the need to manage rare diseases once diagnosed, including any familial risk of inherited conditions.

For the NSW health system to remain responsive to this rapidly evolving healthcare field, new discoveries with patient benefit must be translated into practice. Translating Genomics Research into Clinical Practice' provides a six-step cycle to help researchers plan and conduct genomics research with real-world application. The framework guides alignment between policymakers, clinicians and funders and between government, industry and regulators so implementation barriers are addressed collectively.

2. Enabling systems and infrastructure to support access and integration

Successful integration of clinical genomics into mainstream healthcare requires systems and infrastructure that integrate with clinical workflows and meet consumer needs. Two key NSW infrastructure pieces are the state-wide genomics testing facility provided through the State Pathology Service, and the NSW Health Statewide Biobank. Together, these provide a scalable and affordable model for clinically accredited genomic testing, and for storage of tissue, tumour, DNA and blood samples for use in health and medical research. Services will be enabled by streamlined digital processes for consent, ordering, tracking and reporting of genomic tests, underpinned by robust standards and governance.

3. Upskilling the workforce

Historically, genetic testing has been facilitated by clinical genetics services. However, as clinical genomics mainstreams, skills are needed across the health workforce. The NSW Genomics Strategy is enhancing genomic knowledge, skills and capabilities of the NSW health workforce to ensure appropriate interpretation and use of genomic information in patient care. The NSW Centre for Genetics Education' provides tools and practical implementation guides, helping clinicians and consumers navigate processes for genomic test selection and consent. Training for new genomic workforce functions is also being developed, along with rare disease information for families and clinicians.

The transformative potential of genomics comes with challenges. Our current framing of value requires test results to be linked with clinically actionable outcomes and available treatments that improve health outcomes. Many conditions identified with genomics may have no treatment, or the precision therapy may be very high cost. Discovery of variants...
of known and unknown significance may also cause psychological distress. Ethical, legal and socio-cultural dimensions also require serious consideration. Executive support and cross-functional governance involving multiple agencies, clinical and research partners and consumers are critical to address challenges and barriers, make judicious investment decisions and leverage existing capabilities. Ongoing consumer partnership is essential to maintain public trust and confidence.

Genomics and precision medicine are blurring the line between research and clinical practice. A key part of the strategy is supporting a robust omics translational research pipeline that integrates with clinical care. With only small patient numbers involved, investment in precision therapies is increasingly reliant on a combination of research and real-world evidence. Approaches to research and clinical governance, funding and reimbursement need to keep pace with such innovation.

Genomic data acquisition is increasing rapidly. NSW is working to understand the genomic data-sharing landscape, using insights to build data systems and governance for appropriate storage, sharing and use of data. This strategy positions NSW as a partner to test approaches to data sharing between jurisdictions and agencies, creating models that can be applied nationally.

Genomic information is already improving health outcomes in cancer and some rare diseases, but there is a way to go before precision medicine is fully realised. The NSW Genomics Strategy is one of a range of national and international genomic strategies. With a holistic approach that considers access, infrastructure and workforce in a real-world context, NSW aims to harness potential benefits of genomics at all levels of the healthcare system to benefit the NSW population.

References