



Research Platform 02

# Genomics and bioinformatics

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## Defining genomics and bioinformatics

The human genome is our blueprint for life. It contains all the information needed for us to grow and develop, consisting of 3.2 billion 'letters' of DNA and over 20,000 'chapters' in the form of individual genes. Genomic medicine seeks to read and analyse our genome to gain insights into how each of us is different, as well as understand the causes of malfunction and disease. Bioinformatics combines mathematics, statistics and computer science to solve complex biological problems, including analysing the vast amounts of data produced by genome sequencers.

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**We are entering an age of 'precision medicine', where genomics will help personalise therapies for each individual, dramatically improving our ability to prevent and treat disease.**

Our growing understanding of the human genome is driving a new age of genomic medicine that promises to fundamentally change the way we provide health care at every level. Genomic medicine is giving us a deeper understanding of rare diseases, such as congenital deafness or brain malformations, as well as common diseases including cancers and cardiovascular disease. Genomic tests can also reveal an individual's risk of developing a disease later in life and can help guide preventative strategies that will help them avoid ill health, dramatically reducing the burden on the health care system.

Over 900 Victorian patients have already experienced the benefits of these new genomics approaches through the Melbourne Genomics Demonstration Project. For example, 100 children with

a range of early-onset childhood conditions received genomic tests alongside traditional diagnostic approaches. The outcomes were impressive. When the genomic tests were used, the children were five times more likely to receive an accurate diagnosis, they received far more effective treatment and the overall cost per patient was around one-quarter of traditional diagnostic approaches. Even this small demonstration vividly shows that the health and economic benefits of integrating genomics across the entire health care system would be profound.

Another application of genomic medicine is to provide precision or individualised treatments by tailoring them to someone's specific genetic makeup. For example, many patients currently need to try multiple drugs or therapies until they discover the one that works best for them. This exposes the patient to unnecessary and potentially harmful side effects and also wastes money and resources on ineffective treatments. By understanding a patient's unique genetic makeup, clinicians can choose the most effective drug for that specific individual. In future, they'll be able to manipulate a patient's genes directly or in combination with stem cell therapies to help treat or prevent a wide range of conditions.

The long-term goal of genomic medicine is to have genomics integrated into every level of health care. This will mean getting your genome sequenced will be as simple and inexpensive as having an X-ray and the results can be analysed and discussed with your GP, specialist or genetic counsellor so they can provide the best possible care. This means that preventative strategies can be applied early, staving off serious disease later in life. The incorporation of precision medicine along with the ability to predict and prevent disease will transform health care, improving results and drastically reducing costs.

## Current strengths and opportunities

### Strengthening alliances

The Melbourne Biomedical Precinct is home to international experts in genomic medicine that are leading the way in the integration of genomic medicine into healthcare. The Melbourne Genomics Health Alliance (MGHA) is a collaboration between 10 partners, including research institutes, hospitals and universities across the state. MGHA has received \$25 million from State Government over four years and each partner contributes \$1M equating to a \$35 million investment.

Melbourne Genomics Health Alliance is a key member of Australian Genomics, a consortium of 80 centres nationally, led from the Murdoch Children's Research Institute. Australian Genomics has received \$25 million from NHMRC and is leveraging over \$125 million of investment across the country, to pilot and implement Australia's national diagnostic and translational genomics framework, reduce waste to stem healthcare costs and build a skilled genomic literate workforce.

### Disruptive technology

Genomic medicine will be highly disruptive to the health care industry and more effort is required to integrate genomics into health care at all levels. A key element of the Australian Genomics work plan is to help align national and state policy and legislation about the use and availability of genomic information.

### Expanding sequencing capability

The Australian Genomics Research Facility (AGRF) partnered with The University of Melbourne, Walter and Eliza Hall Institute, Murdoch Children's Research Institute and Peter MacCallum Cancer Centre to launch the Genomics Innovation Hub in 2016, significantly boosting the genome sequencing capacity available to Melbourne clinicians and researchers and providing the opportunity to test and rapidly adopt the latest technologies. AGRF and The University of Melbourne have also purchased three NovaSeq 6000 platforms to further enhance the Melbourne Biomedical Precinct's sequencing capability.

Within 10 years, anyone with diseases with known genetic causes will be able to access the latest genomic technologies, along with all cancer patients and family members deemed at-risk.

### Proven expertise

The Melbourne Biomedical Precinct already has proven expertise and capacity in genomic sequencing and bioinformatics, with many of Australia's leading researchers working for Precinct Partners. The Melbourne Biomedical Precinct is also home to the Bioinformatics Consulting Core, located at Peter Mac, which provides essential data analysis for researchers and clinicians. The Victorian Centre for Functional Genomics at Peter Mac is spearheading the next phase of genomics, working to understand the function of the 20,000 human genes and how they interact to cause disease. There are other types of 'omics' than genomics and the Melbourne Biomedical Precinct has expertise in these areas too, including proteomics and metabolomics, for which the Bio21 Institute is the national hub.

## Future opportunities

### Genomics in precision health

The clinical and research workforce in the Melbourne Biomedical Precinct needs to grow significantly to capitalise on the opportunity of genomics and precision health. Opportunities for professionals to move between research and clinical practice should be fostered and supported.

### Information leadership

Building genomics and bioinformatics infrastructure and expertise in the Melbourne Biomedical Precinct is vital to analyse and gain insights from the data produced by the latest genome sequencing technology. Addressing capacity to store genomic and bioinformatic information is essential to maximise the value and impact of genomic sequencing.

### International partnerships

The leaders of Melbourne Genomics Health Alliance and Australian Genomics play a lead role in the Global Alliance for Genomics and Health, a consortium of over 500 institutions across 46 countries that is developing international standards and frameworks for data sharing and efficient and equitable translation of genomic research on a global scale. Our genomics initiatives will formalise key partnerships with Genomics England, Genome Canada and the US Precision Medicine Initiative - opening up a world of opportunity to develop novel diagnostic and therapeutic approaches, further enhancing our research capacity.

### Opportunities for industry partnership

Precision medicine is currently one of the fastest-growing segments of the health industry, underpinning the development of novel diagnostic approaches, capacity-building in clinical informatics, bioinformatics and bio-specimen banks, and driving drug discovery and development for common and rare diseases.