

Symposium Description (250 words max.)

Symposium Lead (name and affiliation): Associate Professor Louisa Gordon, QIMR Berghofer Medical Research Institute

Rationale for Symposium: Genomic medicine is a new medical discipline that involves using genomic information about an individual as part of their clinical care. Genomics is expected to be widely-adopted in many areas of our health system, across all patient groups and settings. However, evaluations of clinical genomics are sorely lacking and there is an urgent need to evaluate clinical applications of genomic sequencing to determine their value to society. There are very important ethical, legal and social issues associated with genomic testing which highlight issues of equity. The opportunity cost of adopting expensive next-generation sequencing tests when there is no shortage of other urgent public health investment needs is also high. Weighing the pros and cons of the initial testing and subsequent flow-on health care decisions and associated costs has significant challenges in economic evaluation. This symposium will highlight the evaluation experiences of the speakers who are active members of the Australian Genomics, Melbourne Genomics and Queensland Genomics alliances and summarise the issues and challenges for health service researchers in future work.

Topic description: Genomic medicine is rapidly being adopted by clinicians demanding to use genomic information for improving care of their patients. There are some significant challenges for program evaluators of this new technology which will be shared and discussed in the symposium.

Presentation one

Authors and affiliations; A/ Professor Louisa Gordon, QIMR Berghofer Medical Research Institute, Population Health Department; Queensland Genomics

Overview: This presentation will introduce genomic medicine and what it means for health services research. Genomic medicine is increasingly used to personalise medical care for patients but it is expensive and there is an urgent need to evaluate clinical applications of genomic sequencing to ensure society recognises their relative value. This presentation will highlight the current evidence base and what health service researchers need to know about this new 'disruptive' technology. There are many challenging issues relating to evaluation and, social, legal and equity issues will also be discussed.

Presentation two

Authors and affiliations Dr Ilias Goranitis, Centre for Health Policy, University of Melbourne; Australian Genomics Health Alliance, Murdoch Children's Research Institute

Overview: Genomic medicine has transformed patient diagnosis, prognosis and management for many conditions. A key challenge in determining the value of genomics is the wide range of health and non-health benefits they generate to patients and their families. Capturing the value of genomic medicine may require a shift in the conventional methods used in programme evaluations. This presentation will discuss the findings of a stated choice experiment designed to capture the value of genomics in children with neurodevelopmental genetic disorders. The presentation will also reflect on how these findings can be used in a cost-benefit analysis to assist programme evaluations.

Presentation three

Authors and affiliations; Thomas Elliott, QIMR Berghofer Medical Research Institute

Overview: Whole genome sequencing (WGS) can rapidly and accurately identify infectious pathogens. We evaluated the clinical and economic impact of WGS availability in containing a large-scale hospital outbreak of *E.coli* using a hybrid simulation model of the hospital environment. Model inputs were determined using microbiology and WGS data, hospital admission databases and local clinical knowledge. Over 5 months, an estimated 197 patients were colonised during the outbreak with 75 patients detected. The total outbreak cost was AU\$460,137 with 6.1% spent on sequencing. Without WGS, the outbreak was estimated to result in 352 colonised patients costing AU\$766,921.

Presentation four

Authors and affiliations; Dr Astrid Rodriguez-Acevedo, QIMR Berghofer Medical Research Institute

Overview: Myeloid cancers include a group of disorders including acute myeloid leukaemia (AML). The 2-year survival rate for AML is very poor at 54%. Existing risk stratification methods including clinical, pathological and single gene mutation testing are still unable to accurately predict disease behaviour in a majority of patients. Genomic sequencing offers additional information for doctors to appropriately treat patients with stem cell transplantation, distinguishing between those patients who need it the most and those who do not. This presentation will discuss the evaluation approach to assess the value of introducing this new technology to patients and treating physicians.