# Personalised medicine possible with real-time integration of genomic and clinical data to inform clinical decision-making

**Maureen Turner1, Alice Johnstone1, Leon Heffer1, Naomi Rafael1, Advisory Group2, Ivan Macciocca2, Natalie Thorne2, Tim Bakker2, Clara Gaff2**

1 BioGrid Australia, Melbourne, Australia, enquiries@biogrid.org.au

2 Melbourne Genomics Health Alliance, Melbourne, Australia, enquiries@melbournegenomics.org.au

## INTRODUCTION

Over 14,000 genes are known to cause or contribute to disease [1] and over 2000 diagnostic genetic tests are now available in clinical practice [2]. Despite widespread use of genomic sequencing in research, there are gaps in our understanding of the performance and provision of genomic sequencing for clinical use. The Melbourne Genomics Health Alliance (the "Alliance") is a collaboration between seven Melbourne-based research and clinical organisations. The Alliance has been established to determine the feasibility, performance and impact of using genomic sequencing as a diagnostic tool with the objective of demonstrating that personalised medicine through targeted genomic analysis is possible.

To provide access to genomic information in the clinical setting requires the integration of the genomic data into the existing clinical framework. In order to prototype the linkage of patient-sourced information with genomic data as part of the demonstration project, the Alliance has partnered with BioGrid Australia, an independent not-for-profit technology partner, to provide a framework and infrastructure for data linkage and sharing. This presentation will focus on how BioGrid Australia's established systems and processes have supported the demonstration project.

## IMPLEMENTATION PROCESS

BioGrid Australia has provided two areas of expertise to the demonstration project, namely data capture and data-linkage, to assist with the demonstration project's objectives of examining the feasibility of using patient entered data linked with clinical and genomic data.

To capture patient-sourced information, The Alliance worked with BioGrid Australia to develop a secure web-based tool. Working with their stakeholder groups, the Alliance defined the requirements for the patient-entered data collection tool, using existing resources where possible. BioGrid Australia then developed, tested and deployed this application for use by the participants in the demonstration project. This tool has been used to capture information about the participants' health, lifestyle and healthcare and has also captured metrics of data entry. These metrics will be used to evaluate the completeness, accuracy and acceptance by the participants.

The demonstration project requires the integration of diverse patient-level health datasets from a number of disparate sources. A significant challenge associated with this activity is ensuring the protection of patient privacy and confidentiality while maintaining data integrity. The Alliance addressed this by utilising BioGrid Australia's established platform for the ethical and secure linkage of clinical, genomic and patient-entered data.

BioGrid Australia's federated data-sharing platform enables real-time integration of record-level data across institutions and jurisdictions. Distributed datasets are connected to BioGrid Australia's data linkage platform creating a single point of access and removing the need to establish and maintain a centralised database. The platform's data access management system ensures that only researchers authorised by the relevant data custodians can access the specified datasets. This approach provides the Alliance with real-time access to clinical, genomic and patient-entered data stored across institutions and jursdictions for their ethically approved research projects.

Another key feature of the BioGrid Australia platform is its replacement of patient identifiers with a Unique Subject Identifier (USI) before health data is released to authorised researchers. The USI allocation process is securely managed by BioGrid Australia and is completed prior to the linkage of health data. Critically, by using technology that creates non-reversible encrypted linkage keys [3], BioGrid Australia's USI enables the matching of patient-level data without requiring the disclosure of personally identifying information. In this way the Alliance is able to link disparate datasets while ensuring the maintenance of patient privacy and confidentiality.

BioGrid Australia also provides access to data analysis and visualisation tools to enable researchers to query and explore the integrated datasets, as well as providing both project wide reporting and integrated views of patient-level data. The benefit of using the existing BioGrid Australia framework and capabilities for the demonstration project is that it allows the Alliance to examine the feasibility of linking clinical, genomic and patient-entered data without having to establish the necessary infrastructure and processes themselves.

## CONCLUSION

This project will allow the Alliance, through its demonstration project, to facilitate the integration of genomic sequencing into clinical practice and provide recommendations for subsequent implementation. A key process for this implementation is the ability to securely link the diverse health datasets to maximize the utility of the existing information to enhance the clinical decision making process. BioGrid Australia has provided a solution for the secure linkage for the health datasets used as part of the demonstration project. This translational analysis and reporting provides the groundwork for further exploration of targeted approaches to personalised disease treatment and exemplifies the role of integration of electronic health records in enhancing the clinical decision making process.

## REFERENCES

1. Online Mendelian Inheritance in Man, from: www.omim.org/statistics/entry, accessed 17 October 2013
2. Centre for Disease Control, from: http://www.cdc.gov/genomics/gtesting/, accessed 18 October 2013
3. GRHANITE™ Health Informatics Unit, from www.grhanite.com , accessed 4 June 2014

## ABOUT THE AUTHOR(S)

### Maureen Turner

Maureen Turner is the CEO of BioGrid Australia and utilises her extensive commercial experience for the organisation's strategic direction and operational management. Maureen has previously held senior management positions in Operations and Commercial Risk, Business Development and Marketing across a number of industry sectors. Maureen is a Fellow of the Australian Institute of Management and a Member of the Australian Institute of Company Directors. She has a MBA from RMIT University.

### Alice Johnstone

As a project officer with BioGrid Australia, Alice is responsible for providing support to a diverse range of clinical and research projects. Alice has a keen interest in statistics and genetics arising from her background in medical research. She holds a PhD in Molecular Biology from the University of Otago.

### Leon Heffer

Leon Heffer is Head, Data Services at BioGrid Australia. Leon's role entails the strategic management of ethics, data linkage, access and analysis, and custom data-collection software development. Leon has a PhD in Neuroscience from the University of Melbourne and has over 10 years' experience in scientific research and research management.

### Naomi Rafael

Naomi Rafael is the Head, Technology and Systems at BioGrid Australia. She holds the degree of Masters of Information Technology from the University of Melbourne. Naomi leads the BioGrid technical team whose activities consist of ICT governance, IT security, data integration, system architecture and administration, software engineering and metadata management.

### Ivan Macciocca

Ivan Macciocca has been seconded from the Victorian Clinical Genetics Services (VCGS), based at the Royal Children's Hospital (RCH), where he worked as a genetic counsellor for 15 years prior to taking his role as clinical project manager at Melbourne Genomics. Ivan has played an integral role in the establishment of new clinical genetics services, including throughout rural Victoria.

He worked in London at the Heart Hospital managing the translation of individual genetic research findings into clinical care. After returning to Australia, his clinical work focused on inherited cardiac disease, where he was involved in

establishing new cardiac genetics clinics at the RCH, Royal Melbourne Hospital, Monash Medical Centre and The Alfred, together with colleagues at each of these sites. He also contributed to the establishment of a new next-generation sequencing test for genes associated with inherited heart conditions offered by the VCGS and completed a Masters thesis on the impact of genetic testing for these conditions.

**Natalie Thorne**

Natalie Thorne is the project manager, clinical bioinformatics and genomics. After a degree majoring in mathematics and statistics and minoring in genetics, Natalie completed her PhD at the Walter and Eliza Hall Institute's Bioinformatics division.

She worked for Cancer Research UK at the Cambridge Research Institute in the computational biology group where she worked on a variety of microarray genomics applications in cancer and analysis of data from emerging genomic technologies. After five years in the UK, she returned to the Walter and Eliza Hall Institute in a senior postdoctoral position in the statistical genetics lab. Her studies utilised high-throughput sequencing technology to discover disease-causing DNA mutations, focusing on projects to improve the use of whole genome sequencing for disease diagnosis.

**Tim Bakker**

Tim Bakker is the project manager, information management and ICT at Melbourne Genomics.

Tim has worked in the IT industry for more than ten years, most recently working as a project manager at The University of Melbourne, in which he led a number of infrastructure and application projects within the central IT unit. Previous experience has included roles in service desk, system administration and IT management.

**Clara Gaff**

Clara is the program leader for Melbourne Genomics. Clara has been involved in the use of genetics and genomics in health care for almost 20 years through roles in genetic counselling, management of genetic services, health professional education, and strategic development in Australia and the UK. She has worked in public health, government, academic and not-for-profit sectors.

Clara has a PhD in molecular genetics, certification in genetic counselling and postgraduate qualifications in health service research and evaluation. She is a member of the National Health and Medical Research Council (NHMRC) Human Genetics Advisory Committee and the Ethics and Social Issues Committee of the Human Genetics Society of Australia.

**SHORT ABSTRACT**

**Background:**
Over 14,000 genes are known to cause or contribute to disease and over 2000 diagnostic genetic tests are now available. Despite widespread use of genomic sequencing in research, there are gaps in our understanding of the performance and provision of genomic sequencing in clinical practice. The Melbourne Genomics Health Alliance, a collaboration between seven Melbourne-based research and clinical organisations, has been established to determine the feasibility, performance and impact of using genomic sequencing as a diagnostic tool with the objective of demonstrating that personalised medicine through targeted genomic analysis is possible.
**Method:**
The Alliance has partnered with BioGrid Australia to enable the linkage of genomic sequencing, clinical treatment and outcome data for this project. BioGrid operates a secure federated data-sharing platform that enables real-time integration of record-level data across institutions and jurisdictions. This platform provides ethical access to data while protecting both privacy and intellectual property and importantly has the capability to provide an integrated view for the clinician as well as project wide reporting.
**Results:**
This project will build an integrated dataset of genetic, clinical and patient sourced information that can be used to evaluate the potential diagnostic value of genomic sequencing in routine clinical practice. This presentation will focus on the data linkage and sharing framework as well as key implementation challenges.
**Conclusion:**
This project will allow the Alliance to facilitate the integration of genomic sequencing into clinical practice, and provide recommendations for subsequent implementation. This translational analysis provides the groundwork for further exploration of targeted approaches to personalised disease treatment.