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

Alice Johnstone\textsuperscript{a}, Leon Heffer\textsuperscript{a}, Naomi Rafael\textsuperscript{a}, Maureen Turner\textsuperscript{a}, The Advisory Group\textsuperscript{b}, Ivan Macciocca\textsuperscript{b}, Natalie Thorne\textsuperscript{b}, Tim Bakker\textsuperscript{b}, Clara Gaff\textsuperscript{b}

\textsuperscript{a}BioGrid Australia
\textsuperscript{b}The Melbourne Genomics Health Alliance
Melbourne Genomics Health Alliance

• Demonstration project

Feasibility and value of genomic sequencing in clinical practice

Five disease areas
- Acute Myeloid Leukaemia
- Charcot-Marie-Tooth
- Hereditary Colorectal Cancer
- Epilepsy
- Syndromes of Childhood

Exome sequencing
- Specific variant information captured for each disease
BioGrid Australia’s Role

• Linkage
  – Multiple hospitals, multiple genomic laboratories, patient information, clinical outcomes

• Enable Patient-entered data collection
  – Creation of web-based collection tool to capture history and lifestyle information
Background

- Established 2003 via Bio21 Cluster Victoria to address difficulties of conducting patient-specific privacy-protected research across institutions and diseases

- Created by clinical researchers to ethically access existing and incorporate new data sources for clinical research

- Not-for-profit company limited by guarantee owned by 28 members (43 institutions)
Data Sharing Considerations and Challenges

- Maintaining patient privacy
- Ethics approval for process and projects
- Data governance framework
- Secure data management
- Authorised data access
- Protection of intellectual property
- Infrastructure capability
Data Ethics, Privacy and Security

- Ethics approval at each participating institution for BioGrid linkage methodology and processes

- Data access controlled and provided
  - ONLY to authorised users (Data Custodian, HREC)
  - ONLY in de-identified form
  - ONLY for ethically approved projects

- Data governance and security
  - Unique subject identifier (linkage key) encrypted
  - Regular external security audits
  - All data queries monitored and logged

- Data custodian retains FULL control over their data
Hereditary Colorectal Cancer

• Colorectal cancer risk is influenced by lifestyle factors
• Extensive information not routinely collected
  – Could electronically captured patient entered data provide this information?
PARTICIPANT HEALTH SURVEY

Integration of genomic sequencing into clinical care

The purpose of the survey is for the researchers to compare information you provide about your cancer and health history to information contained in your medical record and to test the possibility of linking the information you enter to the results of the genomic sequencing test you are having performed as part of the research study "Integration of genomic sequencing into clinical care".

The Royal Melbourne Hospital
# Melbourne Genomics Health Alliance

## PARTICIPANT HEALTH SURVEY

### Personal Information

If you want to take a break, please click **SAVE** before you logout.

<p>| | | | | | | | | |</p>
<table>
<thead>
<tr>
<th></th>
<th></th>
<th></th>
<th></th>
<th></th>
<th></th>
<th></th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Start</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
<td>5</td>
<td>6</td>
<td>7</td>
<td>8</td>
</tr>
</tbody>
</table>

**Summary**

### 1. Demographics

All your information is entered via a secure connection and is treated as confidential.

**First Name**

**Middle Initial**

**Last Name**

**Gender**

- Female
- Male

Select your date of birth using the dropdown lists

- **Day**
- **Month**
- **Year**

**What country were you born in?**

**What country was your father born in?**

**What country was your mother born in?**

**What language do you speak at home?**

**What is your current postcode?**

**What is the level of the highest qualification that you have completed?**
Personal Cancer History

2. What type of tumor do you have?

If you have a Biopsy/Histology report from your doctor please type in the diagnosis as shown on the report.

Start typing the name of the tumor into the text field. You can choose one of the suggested types which will appear or enter another name.

The location is not in the list? Please select ‘Other’ and specify.

First, select the region of your body:

Please select

Next, select the location of the tumor:

Please select
### Smoking & Alcohol History

#### 7. Do you currently smoke?

- Yes
- No but I smoked in the past
- No, I have never smoked

At what age did you start smoking? __________

At what age did you stop smoking? __________

How many cigarettes did you smoke per day? Please select ▼

#### 7a. Do you drink alcohol?

- Yes
- No but I consumed alcohol in the past
- No, I have never drunk alcohol

Number of days on which you previously consumed alcohol in an average week: Please select ▼

Average number of standard drinks consumed per day: (100mL wine, 375mL beer, 30mL spirit) Please select ▼
At Each Institution

UoM NECTAR

AGRF

UoM Centre for Translational Pathology

VLSCI Pipeline

Variant DB

LOCAL RESEARCH REPOSITORY (LRR)

At Each Institution

VPN CONNECTION

BIOGRID AUSTRALIA

BIOGRID RESEARCH FEDERATOR (FDI)
Visualisation

• Registration tracking

• Exome analysis results will be used for several purposes:
  • Inform treating clinician
  • Inform patient of results

• Potential use for further research projects where consent is given
Whole exome sequencing covers the genetic information relating to the sections of DNA that produce protein only.

A pathogenic variant affects the function of the gene product. This variant may, or may not be associated with the condition.

Sample Details:
- Source: Whole blood
- Date DNA extracted: 08/08/14
- Capture Date: 10/08/14
- Sequencing Date: 12/08/14

Whole exome sequencing covers the genetic information relating to the sections of DNA that produce protein only.
Summary

- Melbourne Genomics Health Alliance demonstration project examines the feasibility of clinical integration of genomic information
- BioGrid has provided a solution for data linkage encompassing governance, security and ethical requirements
- Knowledge gained from the demonstration project will be used to provide recommendations for clinical implementation
BioGrid Australia acknowledges its supporters

- BioGrid Australia Members and Collaborators
- National eResearch Collaboration Tools and Resources
- Victorian Cancer Agency, Victorian Department of Health
- Victorian Government Department of Business and Innovation
- Australian Government Department of Innovation, Industry, Science and Research