Senate inquiry into National Cancer Screening Register (NCSR)

Submission

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About Prof Matt Bellgard and the Centre for Comparative Genomics, Murdoch University

Professor Matthew Bellgard is a computer scientist by background with a PhD from the University of Western Australia in the field of artificial intelligence. His scientific work has resulted in developments in both the areas of pairwise sequence alignment and artificial intelligence, bacterial bioinformatics, whole genome analysis and annotation for a range of species, as well as eResearch within a range of disciplines. Major discoveries include vaccine candidates in bacteria and cattle tick, evolutionary mechanisms in bacterial genomes, molecular plant breeding analysis, new computational biology analysis tools and the development of integrated web-based systems for agricultural, biomedical applications and health. Deployed production systems include National data repositories adopting international data standards, global patient registries, hospital biobanks, laboratory information management systems and web-based analytic environments.

Professor Bellgard is the inaugural Director of the Centre for Comparative Genomics (CCG) based at Murdoch University, having previously been the Director of the Centre for Bioinformatics and Biological Computing in the School of Information Technology. Matthew has resided at the European Molecular Biology Laboratory (Heidelberg, Germany), the National Institute of Genetics (Mishima, Japan) and the National Center for Genome Resources (Santa Fe, NM, USA) during the course of his academic career.

The Centre for Comparative Genomics (CCG) is a Western Australian State Government Centre of Excellence at Murdoch University, Western Australia, launched in 2005. The CCG undertakes research in the biomedical and agricultural sciences on themes as diverse as human health, personalised medicine, food security, environmental management, animal/plant genomics,pathogens and viruses. The key to the CCG's ability to operate at the boundaries between these diverse fields lies in its unique mix of capabilities and expertise in the enabling disciplines of bioinformatics, genomics, molecular therapies, software development, super and cloud computing.

The CCG has established a focus in human health with strong capability and capacity in molecular therapies and in building and deploying global patient registries such as clinical and screening registries, as well as hospital biobank eHealth solutions. This has enabled the CCG to

grow to be Western Australia's leading informatics and molecular therapy Centre with a workforce of 25 and an annual budget of over $4 million. The CCG has attracted in excess of $51 million in research funding over its lifetime.

The CCG's interest in the NCSR stems from our experience developing and deploying disease registries within Australia and Internationally. By way of background, relevant CCG experience includes:

- Development of molecular therapies for rare diseases including recent FDA approval for a new drug for a rare disease, Duchenne Muscular Dystrophy[1];
- Development and deployment of the Myotubular and Centronuclear Myopathy Patient Registry, a global registry managed from the UK and operated by TREAT-NMD at Newcastle University, in partnership with the Myotubular Trust[2];
- Development and deployment of the Familial hypercholesterolaemia (FH) Australasia Network, the National FH Registry[3] that also contains an additional Lipoprotein(a) registry (an innovative *registry within registry* paradigm as the two disease conditions can be efficiently screened by the same clinicians);
- Development and deployment of the Australian National Duchenne Muscular Dystrophy (DMD) Registry, governed through the Western Australian Department of Health, has been developed to collate a patient's DMD gene sequence and clinical information about their disease[4];
- Telethon Speech and Hearing, the Pilbara Ear Health Model of Care (MOC)[5], launched on 21 September, 2016, screening decision support registry development underway by the CCG;
- Establishment of an Independent Rare Disease Registry (IRDR) through detailed interactions with clinicians, patient advocates and big pharma. Designed and implemented a deployable IRDR for Gaucher disease as a model for post-authorisation assessment for orphan drugs[6].

Response to Senate inquiry into National Cancer Screening Register (NCSR)

- I commend the Government for this initiative and completely agree with need for the establishment and ongoing maintenance of cancer-screening registers (I will refer to them now as 'registries')
- Many existing disease registries are driven and funded by community-based patient advocacy groups and not by government policy;
    - In this context, end-user and stakeholder engagement is critical early on in the development of registries;
- Registry requirements evolve over time and have defined key criteria for the development of robust and sustainable registry implementations[7];

[1] http://www.abc.net.au/news/2016-09-20/fda-approves-muscular-dystrophy-drug-eteplirsin/7861024
[2] http://mtmcnmregistry.org/
[3] http://www.athero.org.au/fh/patients/fh-registry/
[4] https://nmdregistry.com.au/dmd/
[5] http://www.tsh.org.au/modelofcare
[6] https://www.rarevoices.org.au/news/3104/an-independent-rare-disease-registry-an-enabling-technology
[7] *Bellgard et al.*, Dispelling myths about rare disease registry system development, https://scfbm.biomedcentral.com/articles/10.1186/1751-0473-8-21

- Governance and compliance arrangements are paramount. Issues that need to be resolved include the need to establish a: i) Governance Access Committee with an appropriate Terms of Reference; ii) Data Sharing Policies; iii) Request for Data Access Policies; and iv) Registry Operations Committee with appropriate Terms of Reference, at a minimum;
- Consideration should be given as to whether Australia should review relevant legislation and guidelines from other jurisdictions such as the US Health Insurance Portability and Accountability Act and EU data privacy provisions;
- Consideration should be given as to how the NCSR investment can be leveraged for other cancers, chronic and rare diseases. There are over 6,000 rare disease alone. Technological innovation, effective strategies for end-user engagement, compliance and governance arrangements, as well as interoperability with other Federal initiatives such as MyHealthRecord[8] should be leveraged back to the community;
- The NCSR investment should form part of a broader national strategy for disease registries. For example, the development of a **National Disease Registry Strategy (NDRS)**
- The development of the **NDRS** would mean any Federal Government investment in registries could be leveraged by other initiatives at both the Federal and State levels; NDRS would encompass: chronic diseases, common inherited diseases; rare diseases; as well as health services for chronic disorders;
- Within the NDRS, Centres of Research Excellence and Innovation should be considered to be formed so that Federal initiatives such NCSR can:
    - **Align to** other important national initiatives such as the NHMRC's Advanced Health Research and Translation Centres[9] that are recognised to have the potential to: i) increase the cost-effectiveness of health care by identifying, testing and introducing systems of care, procedures and devices that are most effective; ii) restrain cost increases by identifying and eliminating those process, procedures and treatments that are ineffective or less cost-effective; iii) provide strong and more effective dissemination of information towards achieving improvements; iv) provide a more powerful base for clinical trials to the benefit of patients and our clinical trials industry; and v) provide much greater efficiency in the use of resources – clinical, community, research through rationalisation, avoiding duplication, and gaining critical mass efficiencies;
    - **Inform** studies such as Australian Commission on Safety and Quality in Health Care: Exploring Healthcare Variation in Australia: Analyses Resulting from an OECD Study[10] which recognises that systematic identification, interpretation and response to unwarranted variation requires a 'self-improving' health care system that combines: i) the accurate collection of treatment and outcome data; ii) the capacity to analyse these data to provide meaningful feedback to clinicians and providers; and iii) the capacity to undertake robust clinical trials that provide definitive answers to important clinical questions that arise from understanding these variations;

[8] https://myhealthrecord.gov.au/internet/mhr/publishing.nsf/content/home
[9] https://www.nhmrc.gov.au/research/advanced-health-research-and-translation-centres
[10]https://safetyandquality.gov.au/wp-content/uploads/2014/05/Exploring-Healthcare-Variation-in-Australia-Analyses-Resulting-from-an-OECD-Study.pdf

- **Be integral to** the Government's Digital Transformation[11], Innovation[12] Agendas and Cyber Security Strategy[13] supporting and enabling innovative research and development working with start-ups, big pharma, ICT Vendors and the health sector;
- **Contribute to** proposals that aim to address gaps in Australia's health system. One such pressing challenge identified is: *The vision of clinical research that drives continuous improvements in health care delivery is unrealised due to challenges in the volume, variety and veracity of health data; which is further compounded by an inability to conduct Internet scale analytics on siloed health data in a timely manner to optimise the patient journey;*[14]
- **Address** specific health challenges that are faced by indigenous Australians. As such, any significant national health investment or strategy should include initiatives that aim to address these challenges;
- **Align to** both the National Collaborative Research Infrastructure Strategy Roadmap[15] and Professor Anne Kelso's (NHMRC CEO) Medical Research for the 21st Century Vision that has clinical registries and biobanks as core research infrastructure for future national medical research.

Summary

I commend the government on this initiative that represents a significant investment in the improvement of health care for Australians. There is an opportunity for this initiative to form an integral part of a broader **National Disease Registry Strategy.** Within this context I believe it is vital that investments such as the NCSR are developed and aligned with:

- National Collaborative Research Infrastructure Strategy Roadmap
- Professor Anne Kelso's (NHMRC CEO) Medical Research for the 21st Century Vision
- NHMRC's Advanced Health Research and Translation Centres
- Federal Government Digital Transformation Agenda
- Federal Government Innovation and Science Agenda
- Medical Research Future Fund
- Federal Government strategies and policies governing ICT procurement[16]
- Federal Government Cyber Security Strategy
- Existing Disease Registry activities

I thank the Committee members for your consideration.

[11] https://www.dto.gov.au/
[12] http://www.innovation.gov.au/
[13] https://cybersecuritystrategy.dpmc.gov.au/
[14] Medical Research Future Fund 2016 Request for submissions to Australian Medical Research and Innovation Two Year Priorities: submission by Prof M Bellgard, **Data science and the patient journey: Evidence-based research for continuous improvement in healthcare delivery**, *available upon request*.
[15] https://www.education.gov.au/2016-national-research-infrastructure-roadmap
[16] https://www.finance.gov.au/policy-guides-procurement/whole-of-government-ict-policies/