Australian Commission on Safety and Quality in Health Care Framework for Clinical Quality Registries
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Thank you for the opportunity for Australian Genomics to provide a submission to the consultation on Australia's Framework for Clinical Quality Registries (CQRs) 2nd addition.

The purpose of CQRs is to maximise the use of data in supporting health service delivery to improve patient outcomes by "achieving national reporting and the return of information to patients, clinicians, health service providers, health insurers, governments and the community on the appropriateness and effectiveness of health care in high-priority clinical conditions, medical devices, therapies and interventions". We acknowledge that some of the issues raised in our submission relate to aspects of the implementation of CQRs periphery to this specific consultation but believe the issues we raise are time critical and are worthy of consideration in a broader context.

About Australian Genomics
Australian Genomics is an Australian Government initiative supporting genomic research and its translation into clinical practice. Through broad engagement and a national collaborative approach, it achieves two key objectives: to improve efficiency, reach and timeliness of genomic research projects, and to support Commonwealth State and Territory health departments in the implementation of genomics research outcomes by refining and communicating evidence to inform policy development.

Australian Genomics engages with current and emerging government policy and priorities to identify gaps and opportunities, to support policy and action for integrating genomic technologies into the health system. By interfacing with consumers, governments, industry and global genomics initiatives, Australian Genomics drives change and growth in the sector.

Consultation on CQRs
Priority areas for data collection
While the prioritisation of areas for data collection was done systematically and with transparency in the Prioritised List of Clinical Domains for Clinical Quality Registry Development Report, rare diseases have been overlooked as a priority area for collection of data in CQRs. While individually rare, there are more than 7,000 rare diseases that are life threatening or chronically debilitating. Rare diseases affect around 2 million people (or 8%) of the Australian population[1]. A study in Western Australia showed that rare diseases accounted for more than 10% of hospital admissions and cost nearly $400

[1] Australian Government. What we're doing about rare disease. 7th November 2022.

million in 2010[2]. In the UK, economic analyses showed a total hospital cost of GBP 3.4 billion in the 10 years prior to a rare diagnosis, or GBP 13,000 per person[3].

There is a need for better rare disease data collection in Australia and development of CQRs represents an opportunity to do this. The Australian National Strategic Action Plan for Rare Diseases 2020[4] reported that data for most rare diseases is not captured. Under the Plan's Research and Data Pillar it calls for more systematic, coordinated data collection through implementation of registries. This will improve monitoring, increase knowledge about rare diseases, and inform clinical practice, research and health service planning.

The UK Rare Disease Plan 2022 aims to improve national disease registration to achieve one of the largest population-based rare disease cohorts in the world. The plan includes standardisation of the minimum core data set and inclusion criteria, coding, and routine analysis, as well as method development for rare disease registration. Rare disease registries will be interoperable with UK's genomic datasets. Rare disease registries are also recognised for their role in identifying health inequalities and supporting clinical research.

Cancer registries are also important but there is better coverage of cancer incidence and screening registries in Australia. Despite fragmentation in approaches, data collection and use is far more advanced than for rare diseases.

Leveraging clinical quality registries for clinical trials and other research

Given the identified unmet need for rare diseases registries in Australia by the National Strategic Action Plan for Rare Diseases[4], the national CQR framework could be applied. This would have the dual benefit of potential for leverage for identifying patients for recruitment into clinical trials and other research. This is an important avenue of access to innovative diagnostic and treatment options, such as personalised, precision therapies. These opportunities are often inequitably applied and available to those who "lucked in" to a clinic with access to such research.

A recent Perspective article in the Medical Journal of Australia addresses this issue and the many advantages of registry-based trials. These include access to baseline and/or outcome data; more cost-effective set up; less duplication; access to PROMs and PREMs; standardised data collection, better representation across the population and application of better trial methodologies[5]. Despite

[2] Walker et al., The collective impact of rare diseases in Western Australia: an estimate using a population-based cohort. Genet Med **19**, 546–552 (2017). https://doi.org/10.1038/gim.2016.143

[3] Mendelian. 2018. A preliminary assessment of the potential impact of rare diseases on the NHS.

[4] Rare Voices Australia. 2020. Australian National Strategic Action Plan for Rare Diseases

[5] Ahern et al., 2022. Realising the potential: leveraging clinical quality registries for real world clinical research. https://doi.org/10.5694/mja2.51443

this, they report that only one trial using CQRs was identified in Australia between 1996-2017. This represents a missed opportunity and should provide the basis for further infrastructural investment and development. This could also serve to promote hospitals as thriving clinical trials and research environments (re ACSQHC National Clinical Trials Governance Framework).

Australian Genomics urges ACSQHC to keep the value of CQRs to clinical trials and research on its agenda as the Framework is implemented. Integrating CQRs and clinical trials also provides opportunities to assess the long-term quality and effectiveness of such interventions.

A rapidly expanding scope for clinical registries

The Framework should consider future demands on CQRs, with the emergence of novel health technologies and interventions: diagnostic, predictive and therapeutic.

With an increasing number of cancer and heritable disease-related cell and gene therapies in the pipeline, the scope and capacity of CQRs should be pre-emptively optimised. For example, the FDA has mandated 15-year follow-up for all gene therapies[6], and the life cycle of therapy development and monitoring will likely need to be matched for the patient life cycle, including the need for longer duration of genomic data storage. This will mean identifying the registry needs for disease-specific, therapy class-specific, or a combination of the two, depending on setting, and longevity of follow up.

As genomic technologies move from diagnostic testing to predictive and prognostic analysis, population-scale genomic screening will soon be available to Australians. Approaches for expanded reproductive carrier testing will need to include means to store results and outcomes and reanalyse data when people recouple; genomic newborn bloodspot screening results, interventions and longitudinal outcomes will need to be recorded longitudinally; and genetic risk for adult-onset conditions (e.g. familial hypercholesterolaemia and high penetrance cancers) will also need to be captured in national CQRs to inform screening schedules and preventative interventions.

The future-focussed guidance provided by the Framework 2nd edition would be strengthened by consideration of the CQR needs of such emerging health technologies.

Good quality, well designed CQRs will be fundamental to achieve a learning health system for Australia and the Commission has an important role in this. We believe the issues raised in our response will be important scope for CQRs, now and in the future.

[6] Food and Drug Administration. Long Term Follow-Up After Administration of Human Gene Therapy Products Guidance for Industry. January 2020.