# Rare Voices Australia Submission: Australian Commission on Safety and Quality in Health Care's Consultation on the Framework for Australian clinical quality registries Second Edition

## Executive Summary

Rare Voices Australia (RVA) welcomes the opportunity to provide a submission to the Australian Commission on Safety and Quality in Health Care's (the Commission) Framework for Australian clinical quality registries Second Edition (the Framework 2nd edition). RVA is the national peak body for the estimated two million Australians living with a rare disease.

Herein, RVA provides a rare disease perspective to relevant areas of the Framework 2nd edition that should be amended to enable the development of much-needed CQRs for rare disease. This perspective is guided by, and aligns with, the priorities, actions and implementation steps of the Australian Government's National Strategic Action Plan for Rare Diseases (the Action Plan)[1]. RVA led the collaborative development of the Action Plan, which was informed by extensive multistakeholder consultation. The Action Plan was launched in February 2020 with bipartisan support, and RVA is now leading its collaborative implementation. The Action Plan provides guidance and direction around key goals and priorities of the rare disease sector in Australia and includes a strong patient voice.

**The need for Clinical Quality Rare Disease Registries**

In rare disease, low prevalence, high burden of disease, high levels of unmet need and variations in care, met with limited treatment options, make registries vital instruments for informing better outcomes. Nonetheless, in Australia, data for most rare diseases is not captured in either health information systems or registries,[2] and there is no coordinated strategy to collect, measure, build and translate data that does exist.

Multiple research papers and stakeholder consultation processes in Australia have identified the need for a national, coordinated, and systematic approach to the collection and use of rare diseases data, including registries.[1] Priority 3.1 of the Action Plan—*Enable coordinated and collaborative data collection to facilitate the monitoring and cumulative knowledge of rare diseases, informing care management, research and health system planning*—directly addresses this gap. It is vital that the Framework 2nd Edition can be leveraged to support the realisation of this priority for rare diseases. It must consider and address the unique barriers to setting up registries for rare disease, including low disease prevalence, and limited data and knowledge.

RVA commends the efforts of the Commission to ensure multistakeholder involvement in the consultation for the Framework 2nd Edition. CQR for rare disease are vital to informing improvements in health systems for better health and wellbeing outcomes for Australians living with a rare disease. RVA strongly advises that revisions to the National Framework for CQR effectively consider and enable the development of CQR for rare diseases.

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## Rare disease considerations for the Framework Second Edition - Enabling CQRs for rare disease

Overall, the Framework 2nd Edition was presented clearly and in simple language. As the national peak body for Australians living with a rare disease, and not an organisation directly involved in developing CQRs, RVA does not have expertise to comment on the information required for implementation of the Framework 2nd Edition. Nor is RVA in a position to comment on the content in Attachments 1 and 2. RVA can offer a rare disease perspective to ensure any amendments to the Framework 2nd Edition enable the much-needed development of CQRs for rare disease. The following sections address specific areas where the Framework 2nd Edition should be made more fit-for-purpose for rare disease.

### Strategic Principles (1.1) and Prioritisation Criteria for National CQRs

There are several barriers to potential development of CQRs for rare disease across some of the Strategic Principles and Prioritisation Criteria for national CQRs.

There are over 7000 known rare diseases and an estimated 2 million Australians living with a rare disease. However, limited data is a common feature of rare diseases. Currently in Australia, rare diseases are not accurately coded at the point of diagnosis, and compounding this barrier to data collection is the growing number of Australians living with an undiagnosed rare disease. These inherent complexities in rare disease are a barrier to development of clinical quality rare disease registries, which are, more often than not, going to face challenges in meeting bullet point 1 under Principle 4:

> *'…reporting on the national burden of disease and the cost of therapeutic interventions using data:*
>
> - *Collected at the point of care on an entire population of interest'*

Similar barriers to developing clinical quality rare disease registries exist in the Prioritisation Criteria. Specifically, it will be difficult in rare disease to meet points under Section 2, Feasibility, outlined in Table 1.

**Table 1. Potential barriers to meeting feasibility criteria when developing CQRs for rare disease**

| Feasibility Criteria | How this is barrier to the development of clinical quality rare disease registries |
|---|---|
| ***2.2 The relevant clinical population can be captured*** | Rare diseases are not routinely coded in health systems making it difficult to identify patients. |
| ***2.3 The clinical condition or event can be systematically recognised*** | Often, rare diseases are complex, multisystem disorders managed by a range of health professionals and there are very few national guidelines on diagnosis and management. This means people living with the same rare disease may undergo different diagnostic journeys and be managed very differently depending on their clinical team and even their postcode. These issues make systematic recognition a challenge. |
| ***2.7 An entire population with a chronic condition or disease, or who have undergone an acute event can be captured*** | As above |
| ***2.8 There is a suitable data source*** | Existing rare disease datasets are often heterogenous and incomplete. The lack of consistent use of rare disease codes is a challenge to identifying suitable data sources in healthcare systems |
| **2.9 Clinically meaningful performance indicators can be defined** | There are few performance indicators or validated tools for measuring clinical care outcomes for rare disease. Such indicators may exist for a fraction of the over 7000 known rare diseases. |

The current Strategic Principles and Prioritisation Criteria for CQRs, may prevent both the development and future accreditation of nationally recognised clinical quality datasets for rare disease. With rare disease data collection and use already limited, these additional barriers could delay or prevent evidence-based improvements in diagnosis, care, support, and access to novel health technologies for the over 2 million Australians living with a rare disease.

## Operating Principles (1.2) for national CQRs

### Model 1 in Figure 3

Model 1 in Figure 3 does not list the different stakeholders involved, as in the other two diagrams for Models 2 and 3. The reason for this is unclear. RVA recommends highlighting stakeholders, including consumers, across all diagrams to avoid confusion (at a glance) and ensure Models 2 and 3 are not viewed by the audience as superior to Model 1.

### 1.2.1 Attributes of clinical quality registries

Acknowledging that, under the Framework 2nd Edition, perhaps not all registries will be required to have every attribute listed in section 1.2, it is important to note that ***Attribute 5 'Outcome determination should be undertaken at a time when the clinical condition has stabilised or an***

***event, such as death, has occurred and the outcome can therefore be reasonably ascertained'*** is not sufficient nor applicable in the context of rare disease. Many people living with a rare disease never stabilise. Rather, rare diseases are often chronic, progressive and life-limiting. To this end, it is not helpful to only measure outcomes related to stabilisation of, or death resulting from, rare disease. For people living with rare disease, it is important to measure outcomes at various points in the rare disease journey. For example, at diagnosis, after beginning a new or novel treatment (even if the patient remains undiagnosed), and through to palliative care, which can be required earlier in the trajectory of life-threatening conditions, including many rare diseases.

**Attribute 6 'Australian national CQRs should seek to ensure there is complete data collected from the entire eligible population'** is also impossible to achieve for rare disease registries in Australia, as existing coding systems are not sufficient or routinely implemented to capture rare disease. Until such a time when people living with a rare disease, or an undiagnosed rare disease, are coded and made visible in health care systems and other systems, it will remain difficult to ensure complete datasets covering the entire eligible rare disease population.

Under Pillar 3, Research and Data, Priority 3.1 of The National Strategic Action Plan for Rare Diseases calls the sector to *'enable coordinated and collaborative data collection to facilitate the monitoring and cumulative knowledge of rare diseases, informing care management research and health system planning.'* RVA is committed to advocating for the realisation of this Priority and its associated actions. RVA recommends that the Framework 2nd Edition considers building flexibility into its Strategic Actions, Prioritisation Criteria and the aforementioned attributes to enable the development of clinical quality registries for rare disease.

### 1.2.3 Data elements, Point 12.

It is important to ensure the information under this heading is articulated in lay terms at the point of patient consent.

**1.2.3 Data elements, Point 15.**

Objective measures or the use of standardised and validated tools may not be possible for rare diseases, as these do not always exist due to difficulties in developing such tools with very limited numbers of patients. The same issue applies to Point 16 regarding validated PROMs tools. RVA is aware this is a growing space and work is being done to establish such tools for some rare diseases. A broad standardised tool that can be used for all rare diseases may be possible through drawing on commonalities.

**1.2.10. Information output, Point 36.**

Regarding statistically meaningful reporting from rare disease datasets, reaching a representative proportion of data by conventional definitions for a statistically significant sample is problematic for rare disease where numbers are low. This is even more difficult for ultra rare diseases. In such cases, considerations around collecting or integrating international datasets are needed, as well as employing statistical methods that can support meaningful reporting on small datasets. RVA recommends the Framework 2nd Edition acknowledges and allows for these nuances of rare disease datasets, as the high burden of living with a rare disease increases the utility of even small datasets for driving better and more cost-effective care.

## 1.3 Governance

RVA strongly supports stringent and consistent governance structures for national CQR that emphasise the privacy, security and informed consent of patients. Strong governance across these areas will improve patient trust, increasing participation in registries.

*Minor note:* Part of bullet point 5 under 1.3.1 'Governing body function', is repeated in bullet point 9. Please ignore this comment if this repetition is intentional.

**1.3.4 Management Committee**

Consumer expertise is not explicitly mentioned in this section, although it is mentioned later under 'Patients and consumers'. RVA suggests inclusion of consumer representatives on the Management Committee. The Management Committee's responsibilities around CQR design, development of a minimum dataset and methods for data collection would all benefit from consumer input. Consumer insights would ensure that data collected in a minimum dataset are meaningful to their communities, thus improving utility of the data. Consumer expertise on this committee would further ensure CQR design and mechanisms for data collection are broadly acceptable to, or approved by, their communities, in turn supporting recruitment.

RVA strongly recommends the CQR workforce include clear reference to consumers. Involvement of consumer expertise should be sustainable and supported, including considerations for, and direct reference to, their remuneration in the Framework 2nd Edition.

**Figure 5 CQR Governance Model.**

It is not clear what the middle row under 'Governance body (legal entity)' is linking to. Is it highlighting the flow of information reported from the governance body and shared systematically through each of those groups to get to the final group 'Contributing hospitals'? Or, is it attempting to show that the governing body is responsible for ensuring all the groups underneath are in compliance with their set of rules for recruiting patients/collecting data etc?

**-END-**

## References


1. Australian Government Department of Health. National Strategic Action Plan for Rare Diseases. Canberra; 2020. 63 p. Available From: https://www.health.gov.au/resources/publications/national-strategic-action-plan-for-rare-diseases
2. Lacaze P, Millis N, Fookes M, Zurynski Y, Jaffe A, Bellgard M, Winship I, McNeil J, Bittles AH. Rare disease registries: A call to action. Int Med J. 2017;47(9):1075–79. Available from: https://doi.org/10.1111/imj.13528

## Appendix A – Relevant excerpts from the National Strategic Action Plan for Rare Diseases

### Priority 3.1

*Enable coordinated and collaborative data collection to facilitate the monitoring and cumulative knowledge of rare diseases, informing care management, research and health system planning.*

#### Action 3.1.1

*Health information systems identify and measure rare diseases and undiagnosed rare diseases.*

**Implementation**

**3.1.1.1.** The Australian Institute of Health and Welfare (AIHW) re-establishes the Australian National Congenital Anomalies Register (NCAR), including rare disease coding (Orphacodes). This will accelerate, extend and nationalise rare disease coding already underway in the Western Australian Register of Developmental Anomalies (WARDA), and contribute to International Classification of Diseases 11th Revision (ICD-11) preparedness.

**3.1.1.2.** Develop a nationally-recognised definition of undiagnosed rare diseases in consultation with relevant experts. Using this definition, provide for an undiagnosed rare disease code in an individual's health record that is compatible with Orphacodes, ICD-11 and other relevant classifications. This code could:

- raise a flag or alert to health professionals when they access the individual's health record (similar to drug allergy alerts), thus prioritising a diagnostic response and;
- support data collection for undiagnosed rare diseases, and hence strategic decision-making, such as service planning.

**3.1.1.3.** Provide for rare disease codes in patient records that are compatible with Orphacodes, ICD-11 and other relevant classifications. This code could:

- raise a flag or alert to health professionals when they access the individual's health records, thus leading to appropriate care that takes into account the rare disease diagnosis and;
- support data collection for rare diseases, and hence strategic decision-making.

**3.1.1.4.** Ensure rare disease and undiagnosed rare disease codes link with a person's Aboriginal and Torres Strait Islander status to allow for culturally appropriate care, and to build evidence of rare disease epidemiology among Aboriginal and Torres Strait Islander people.

### Action 3.1.2

*Undertake broad epidemiological surveillance of rare diseases to support decision-makers to access the information they need to improve the health and wellbeing of Australians living with a rare disease.*

**Implementation**

**3.1.2.1.** Building on existing newborn screening and congenital anomalies data collections, further develop Australia's monitoring of rare diseases and undiagnosed rare diseases. Examples may include and extend beyond:

- newborn and paediatric age ranges; and
- the rare diseases currently captured in newborn screening and congenital anomalies data collections.

**3.1.2.2.** Establish a dedicated Rare Disease Office within the AIHW that publishes periodic national reports on the epidemiology of rare diseases and undiagnosed rare diseases in Australia, including among Aboriginal and Torres Strait Islander people.

### Action 3.1.3

*Improve rare disease data collection and use, including best-practice safe storage, data sharing, custodianship, analysis, reporting and privacy requirements.*

**Implementation**

**3.1.3.1.** Establish a dedicated Rare Disease Office within the AIHW. Included in its remit will be systematic improvements in rare disease data integration and interoperability.

**3.1.3.2.** Publish appropriate data collected through post-market surveillance mechanisms, including under the LSDP to enable better data use and the accumulation of rare diseases knowledge.

## Action 3.1.4

*Develop a national approach to person-centred rare disease registries to support national standards, best practice and minimum data sets.*