rare voices
AUSTRALIA

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Submitted to **Medical Research Future Fund consultation to inform the second Australian Medical Research and Innovation Priorities 2018-2020**
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Introduction

1 What is your name?
Name:
Nicole Millis (on behalf of Rare Voices Australia)

2 Are you affiliated with an organisation?
Yes

3 What kind of organisation do you work for?
Consumer representative, Non-government organisation, Other

4 Are you representing your organisation in making this submission?
Yes

5 What state or territory do you live in?
Victoria

6 Which 2016–2018 MRFF Priorities do you think need further focus? (please select a maximum of three Priorities)
Clinical quality registries

7 How can the 2016–2018 MRFF Priorities you identified in Question 6 be extended or re-emphasised in the 2018–2020 MRFF Priorities?
How can the most important Priority identified in Question 6 be extended or re-emphasised? (max 500 words):
Although Clinical Quality Registries (CQRs) were recognized in the 2016-2018 MRFF Priorities, subsequent MRFF funds have not been specifically allocated towards registries. MRFF funds have been allocated towards clinical trials for rare disease (RDs) and rare cancers, an initiative we wholeheartedly support. However, we believe that high quality registries remain a significant unmet need, and in the particular case of RDs, are essential for to allow effective clinical trials to be undertaken.

Rare Disease Registries (RDRs) is a neglected area that is of critical importance for improving care for the almost 2 million Australians estimated to live with RD. As a sub-set of CQRs, RDRs remain in desperate need of more stable and long-term funding in Australia, and should be re-emphasised as a continuing MRFF priority in 2018-2020. RDRs are historically very difficult to fund via NHMRC or DoH initiatives, yet play a critical role in building a foundation to improve RD research and healthcare. Although the number of rare diseases is large, we believe that individual RD registries could be consolidated in a way that would allow economies of scale.

In Australia RDs are generally defined as disorders with a prevalence of <1/2000. Although each individual RD is by definition infrequent, the combined health burden of all RD in Australia is significant, with an estimated [3] 6% of the total population (or 1/17 people) affected. However, these estimates are limited by a scarcity of robust representative data, due to the lack of specialist RDRs and data collection systems.

The case for Rare Disease Registries:
- RDRs are pivotal for diseases with low prevalence and propensity for variation in treatment and outcome, including diseases with small patient numbers, complex delayed diagnoses, a propensity for variable standards of care and limited treatment options.
- Current lack of RD registries results in uncertainty of basic information including: the number and prevalence of RD in the population; how many patients with a RD are receiving treatment; variation in the quality of care and health outcomes across the health systems; and the overall burden of RD nationally. Without RDRs it will continue to be impossible to ascertain even this basic, yet crucial information for RD in Australia.
- RDRs, if populated with accurate clinical data over extended periods of time, enable far better health service planning and translational research to address current knowledge gaps about RD.
- RDRs act as a central networking point for all stakeholders around a particular RD, including patient advocacy groups, researchers, clinicians, industry and Government.
- RDRs enable clinical trial recruitment and post-marketing drug surveillance, highly timely given the number of new RD drugs becoming available – including Spinraza (nusinersen) for Spinal Muscular Atrophy - recently listed on the PBS at an estimated cost of A$50M+ per year and Orkambi for Cystic Fibrosis, recently recommended by for listing by PBAC.

If you identified a second Priority in Question 6 please explain how it needs to be extended or re-emphasised? (max 500 words):

If you identified a third Priority in Question 6 please explain how it needs to be extended or re-emphasised? (max 500 words):

8 What unaddressed gaps in knowledge, capacity and effort across the healthcare system and research pipeline need to be addressed in the 2018–2020 MRFF Priorities?

Most important gap identified that needs to be addressed in the 2018-2020 MRFF Priorities (max 500 words):
The burden of Rare Diseases (RD) is unacceptably high for patients, families and the community in Australia and needs to be recognised as a national research priority. We support all quality research aimed at improving the detection, prevention, treatment and care of Australians living with RD, and believe that a focus on improving and integrating RD registries will underpin enhanced outcomes for RDs in key translational research areas including, but mot limited to, clinical trials.

At present, the impact of RD remains largely hidden due to inadequate information systems of a healthcare system designed to respond to individual diseases with much larger patient numbers. As covered above, registries are important clinical and research tools and powerful cost-effective instruments to support clinical trials and translational research, and have the potential to improve quality-of-care, quality-of-life and survival. Australia must find a way to better support and fund registries, to address the current gaps – especially in the area of RD.

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RD registries are currently fragmented across individual RDs, with little co-ordination and integration. In their call for a National Rare Disease Framework, Rare Voices Australia have specifically highlighted the need for an integrated approach to registries and data collection to underpin innovative research and clinical trials in RDs across Australia. This is one of 6 key priorities of the RVA framework with the key points listed below.

Priority 3 – Data collection (June 2017)
1. Healthcare coding system to automatically identify and measure RD in Australia
2. Development of evidence-based RD policy based on data that quantifies the collective impact of RD on healthcare, service planning, clinical guidelines and
research.
3. Development of an integrated rare disease registry strategy for Australia

This RVA call is echoed in similar policy and framework documents from a number of International organisations including the European Organisation for Rare Diseases, the National Organization for Rare Disorders, the Canadian Organization for Rare Disorders and the Asia-Pacific Economic Cooperation (APEC). The need for integrated registries to support vital life-saving research is recognized as a priority at the highest levels internationally, and developing an integrated national system in Australia will not only support research nationally, but will align Australia with international goals and enhance collaboration.

If you identified a second gap please explain how it needs to be addressed in the 2018-2020 MRFF Priorities (max 500 words):

If you identified a third gap please explain how it needs to be addressed in the 2018-2020 MRFF Priorities (max 500 words):

9 What specific priority or initiative can address the above gaps?

What specific priority or initiative can address the first gap identified in Question 8? (max 500 words):
The Australian National Alliance of Rare Disease Registries

Rare Voices Australia supports the need for a National RD Registry plan, and has recently led the formation of a National Alliance of Rare Disease Registries. The RDR Alliance is led by an experienced group of RD researchers and clinicians, as a Sub-Committee of the RVA Scientific and Medical Advisory Committee. The Alliance is committed to improving the national landscape for RD patients. Several leading Australian RD registries and support organisations have already joined as members, including the Australian Cystic Fibrosis Data Registry and others. Groups with highly specific registry expertise, knowledge and experience, such as Monash University Registry Science Unit (RSU) and the WA Office of Population Health Genomics have agreed to underpin and support the Alliance, in partnership with RVA.

Members of the National Alliance of RD Registries will work together under a unified umbrella, committing to the following standards and principles:
• Aligned with the guiding principles of RVA
• Share registry best practice, expertise and resources where possible
• Develop and use agreed minimum data sets
• Contribute to the National Operating Principles Document for Rare Disease Registries
• Embrace industry involvement and help to achieve economies of scale
• Promote the collection of patient-centred Quality-of-Life data

- Provide frameworks, tools and assistance for establishing new registries
- Practice inter-operability and consider linking registries in related disease areas
- Remain consistent with international best practices and rare disease networks
- Work towards a nationally recognised process for ethics approval and consent
- Consider future goals, such as a national RD biobank and database

The Alliance puts in place a much-needed cohesive platform for a coordinated national effort to improve RD research and care in Australia. We are inviting many different categories of RD stakeholders to join the Alliance, including existing RD registry operators, individuals and groups interested in starting new RD registries, RD patient groups, government bodies, research organisations and industry.

MRFF support for the Australian National Alliance of Rare Disease Registries would have far-reaching impacts on the RD landscape in Australia, enhancing many aspects of RD research including clinical trials, and ultimately improving outcomes for Australians living with RDs. The fully supported development of an integrated National Alliance for RDRs would align Australia with international initiatives and make Australia a key contributor to international efforts in the RD space.

If you identified a second gap in Question 8 what specific priority or initiative can address this gap? (max 500 words):

If you identified a third gap in Question 8 what specific priority or initiative can address this gap? (max 500 words):

10 What Strategic Platforms (identified in the MRFF Strategy document) would the Priority/ies you identified in Question 8 fall under?
Data and infrastructure, Health services and systems, Capacity and collaboration, Trials and translation

11 How can current research capacity, production and use within the health system be further strengthened through the MRFF? (max 500 words)
Please give us your views:

12 Do you have any additional comments on the Discussion Paper? (max 250 words)
Please give us your feedback on the Discussion Paper:

13 Do you consent to this submission being made public on the MRFF website?
Yes

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