Medical Research Future Fund – Missions Program Evaluation

February 2026

Rare Voices Australia Submission

EXECUTIVE SUMMARY

Rare Voices Australia (RVA), the national peak body for Australians living with a rare disease, welcomes the opportunity to contribute to the review of the Medical Research Future Fund (MRFF) Missions Program.

The MRFF Missions Program, must continue as it has been the most transformative mechanism for rare disease research in Australia. However, this impact has largely been confined to the Genomics Health Futures Mission. To RVA's knowledge, non-oncological rare diseases remain significantly underrepresented across the broader Missions Program.

Approximately two million Australians live with one of more than 7,000 rare diseases. Collectively, rare diseases are not rare. They are chronic, complex and often life-threatening, and fewer than five per cent have an effective treatment. Rare diseases contribute disproportionately to hospitalisation, childhood mortality, lifelong disability and psychosocial burden[1-4]. Internationally, they are recognised as a global health priority.

The Australian Government's National Strategic Action Plan for Rare Diseases[5] (the Action Plan) calls for a coordinated national research strategy to address systemic evidence gaps. The rare disease community has further developed Australia's Top 10 Rare Disease Research Priorities[6] to guide public investment towards areas of greatest unmet need. Despite this clear policy direction, non-oncological rare diseases remain largely invisible within the current MRFF Missions framework.

Targeted rare disease investments, such as genomic newborn bloodspot screening, demonstrate what is possible. MRFF funding has catalysed national consortia, strengthened infrastructure, enabled life-changing diagnoses and positioned Australia as a global genomics leader. But without sustained and dedicated investment in rare disease, Australia risks losing momentum, capability and research talent.

Rare disease research represents one of the clearest examples of unmet need and one of the most powerful drivers of innovation. It has historically served as a proving ground for platforms that now define modern medicine, including precision diagnostics and gene and cell therapies.[7] Investment in rare disease research advances outcomes not only for small populations, but for the broader health system.

Evaluation of the Missions Program presents a generational opportunity to ensure rare diseases—beyond oncology—are recognised proportionately to their burden, complexity and innovation potential, in line with the MRFF's commitment to addressing areas of greatest unmet need.

RVA recommends:

- Establishing a dedicated MRFF Mission for non-oncological rare diseases and appointing a specialist rare disease advisory group to guide mission design and implementation.
- Aligning funding calls with the Action Plan and Australia's Top 10 Rare Disease Research Priorities.
- Embedding policy readiness and translation planning within mission design to support translation readiness
- Improving representative consumer and community involvement
- Ensuring sustained funding through the full disbursement of allocated funds, and greater centralisation and coordination of funding
- Strengthening national coordination, infrastructure and internationally interoperable trial and data systems.

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About RVA

RVA is the national peak body for Australians living with a rare disease, providing a unified voice to advocate for policy, health, disability, and social systems that work for people living with rare diseases. Our person-centred approach brings together key stakeholders across government, research, healthcare, and industry to drive the best possible outcomes for Australians living with a rare disease.

In 2018, the Australian Government commissioned RVA to lead the collaborative development of the National Strategic Action Plan for Rare Diseases (the Action Plan).[5] The Action Plan is the first nationally coordinated effort to address rare diseases in Australia and was informed by an extensive multi-stakeholder consultation process led by RVA. The Minister for Health launched the Action Plan in February 2020, with bipartisan support. RVA continues to work with State and Federal Governments, and all other rare disease stakeholders, in leading collaborative implementation of the Action Plan.

RVA and Research

RVA welcomes genuine partnerships with groups planning or undertaking research that broadly aligns with one or more of the research priorities detailed in the Action Plan. In line with the Research and Data Pillar, RVA supports the need for high quality collaborative research that positively impacts the lives of Australians living with a rare disease.

RVA engages with researchers in several ways, including via the RVA Research Partnerships Program, RVA Scientific and Medical Advisory Committee and the newly formed Australian Rare Diseases Research Network. RVA also has formal links with international research consortia, including the International Rare Disease Research Consortium (IRDiRC) and the European Rare Disease Research Alliance (ERDERA).

The Need to Prioritise Rare Disease Research

There are approximately two million Australians living with a rare disease. People living with a rare disease face common challenges, including difficulty obtaining an accurate diagnosis, lack of treatment options and inadequate, fragmented care.

Rare diseases, like many other chronic diseases, are often serious and progressive. They typically display a high level of symptom complexity and are a significant cause of ongoing health and psycho-social challenges. It has been reported that fewer than 5 per cent of rare diseases have an effective treatment[8].

Rare diseases are recognised internationally as a global health emergency—by the United Nations and World Health Organization—and have been identified by the World Economic Forum and KPMG as 1 of 3 disease domains critical for healthcare sustainability in the Asia–Pacific region.

The burden is significant. In the United States, rare diseases account for two thirds of hospital inpatient costs (USD130 billion annually, versus USD90 billion for common diseases) and nearly half of adult inpatient costs[1]. Comparable figures are evident in Australia. Western Australia health data show only a subset of rare diseases account for 10.5% of inpatient costs system-wide; this extrapolates to AUD400 million per year[2]. For individuals, the annual cost burden is 10 times higher than for common conditions, yet access to tailored treatments can reduce this by over 20%[3]. Mortality is equally stark. Rare diseases cause 6 in 10 childhood deaths[4]—more than cancer and trauma combined—with hospital mortality rates 13 times higher than common conditions[2].

The Missions Program is perfectly positioned to address these pervasive cross-system impacts of rare diseases, aligning with the program's aim to address the big health challenges.

Recommendations for Improving the MRFF Missions Program

The following recommendations are informed by RVA peak body expertise, supported by the RVA Scientific and Medical Advisory Committee (SMAC), and reflect perspectives shared by members of the Australian Rare Disease Research Network (ARDRN).

Recommendation 1. Establish a Dedicated Mission for Non-Oncological Rare Diseases

Through this opportunity to evaluate the MRFF Missions Program Australia has a generational opportunity to position rare diseases as a national research priority, alongside cancer and other more commonly considered disease areas. This approach aligns with the recent prioritisation of rare diseases by the World Economic Forum, fuelled by two critical drivers: (1) profound health inequities, and (2) recognising rare diseases as a critical window into understanding health for all— both common and rare conditions.

Failure to identify rare diseases as a research priority in Australian policy and public research funding risks overlooking one of the greatest drivers of healthcare cost and leaving Australia out of step with international leadership.

Australia is home to a growing number of world-leading rare diseases researchers, clinician-researchers and emerging rare diseases research flagships, centres, and transdisciplinary research teams, making our nation well-positioned to become a global leader in rare diseases research. The expansion of rare disease knowledge is leading the drive behind advances in precision medicine and the development of cell and gene therapies. However, without dedicated funding calls we risk falling behind and losing brilliant minds to other nations or professions.

Despite this, currently, the Missions Programs appears to completely overlook non-oncological rare diseases. For the Missions Program to better respond to rare disease, RVA, RVA SMAC and members of the ARDRN strongly advise expanding the program to include a dedicated mission for non-oncological rare diseases. RVA also strongly recommends the appointment of an expert advisory group for rare diseases to ensure the program respond adequately to rare disease research gaps highlighted in the Action Plan and the Top 10 Rare Disease Research Priorties[6].

Furthermore, the collaborative approach to the Missions Program is critical for rare disease research, where there is a strong need to use resources and expertise wisely and address common challenges for national impact and equity. RVA has welcomed and celebrated the cross-disciplinary rare disease consortia, which have organically formed through targeted calls from the GHFM.

It is also important to highlight that given the small patient populations inherent to rare diseases, international collaboration on rare disease research priorities is essential not optional. Any changes to the Missions Program should work to enable this.

Without a dedicated rare disease call there is a risk to meeting the MRFF's measures of success

Given the limited number of successful rare disease grants to date, the MRFF's measure of success in the MRFF Monitoring Evaluation and Learning Framework (the Framework) to *'increase focus of research on areas of unmet need'* is not being met. Additionally, despite the fact that clinical trials may be the only way for individuals living with a rare disease to access a treatment, to RVA's knowledge very few clinical trials have been funded through the MRFF more broadly, so a similar argument could be made for the Framework's *'clinical trials access'* measure of success.

Notably, although the MRFF performance indicators and measurable outputs mention rare disease, to RVA's knowledge, the Health and Medical Research Office has no way to accurately capture data on rare disease research outputs. This lack of monitoring limits the ability to measure success of the Missions Programs for rare diseases. RVA strongly recommends that the Missions Program formally captures data to enable tracking and measurement of rare disease research funding and impact.

Recommendation 2. Embed Policy Landscape Analysis into Missions Program Design to Support Translation Readiness

In addition to inclusion of a dedicated non-oncological rare disease mission, RVA strongly recommends policy is explicitly recognised and leveraged by researchers and funders as a core enabler of translation. Research builds evidence, but policy delivers equity and impact. The Missions Program should encourage researchers to access and embed policy expertise in their teams, for assessing policy readiness and ensuring environments conducive for translation.

Supporting and resourcing relevant clinician-researcher roles in MRFF Missions Programs is another important mechanism for embedding research into healthcare for effective translation, which is critical to best practice in rare disease health care. More broadly, RVA would like to see greater value placed on, and an expectation for, seeking policy expertise as part of research programs, like the Missions Program, geared towards translation.

Clear examples and evidence of Missions Program-funded projects that perpetuated inequity due to ineffective translation are discussed below, in the section entitled *'The Impact of the MRFF Missions Program on Rare Disease Research'.* Examples of successful outcomes are also highlighted.

Recommendation 3. Improve Representative Consumer and Community Involvement

Another important lever for effective research translation through the Missions Program is to improve how the program involves consumers in research. Below are some recommendations for improving consumer and community involvement from RVA's submission to the revision of the Statement of Consumer and Community involvement in Health and Medical Research:

- Encourage active, early and continuous consumer engagement rom conceptualisation to outcomes, including planning for next steps when projects conclude.
- Provide more tangible examples and set clear expectations for how consumers can be actively and meaningfully changed and acknowledged.
- Prioritise representative consumer involvement that is solution-focused and strengths-based.
- Resource researchers to invest the time required to build respectful, trusted partnerships.
- Implement accountability mechanisms, such as mandatory reporting on consumer involvement and opportunities for direct consumer feedback to the Missions Program team.

Recommendation 4. Provide Explicit Support for Infrastructure, Workforce and Functional Studies—Beyond Clinical Trials and Cohort Studies.

Members of the ARDRN expressed strong concern that current MRFF funding levels, staffing costs, and infrastructure support do not reflect the true cost of conducting high-quality rare disease research. Members expressed the need to strengthening national coordination, infrastructure and internationally interoperable trial and data systems for rare diseases. As mentioned earlier, best practice in rare disease care cannot happen without embedding research and clinical trials.

Recommendations 5. Ensure Sustained Funding Through the Full Disbursement of Allocated Funds, And Greater Centralisation and Coordination of Funding

Surveyed ARDRN members supported greater centralisation and coordination of funding to encourage national collaboration and reduce duplication. They called for better alignment between MRFF and National Health and Medical Research Council (NHMRC) schemes, including pathways for highly ranked NHMRC rare disease applications to be considered for MRFF Mission Program funding. Reducing the number of funding calls while increasing the size of funding pools was seen as critical to lowering administrative burden on both researchers and reviewers. ARDRN members also supported competitive renewal of MRFF-funded programs, particularly in emerging areas such as genomics-based newborn screening, to ensure continuity, innovation and long-term impact.

The Rare Disease Policy Landscape: The National Strategic Action Plan for Rare Diseases and Australia's Top 10 Rare Disease Research Priorities

The MRFF Missions Program aligns with the Action Plan and has been critical to funding research that progresses Action Plan implementation. The Action Plan highlights the gaps in knowledge and evidence across rare diseases, of which there are over 7000 known, and across the research pipeline from fundamental to translational research.

In direct response to Action Plan priority **Priority 3.2: "**Develop a national research strategy for rare diseases to foster, support and drive all types of research for rare diseases, contributing to agreed priorities and systematically addressing gaps", RVA and the Kids Research Institute recently published a priority setting partnership report outlining Australia's top 10 Rare Diseases Research priorities, according the consensus opinions of Australians living with rare diseases, their parents, carers health professionals and community advocates. The report is available via the RVA website:

RVA together with members of RVA SMAC and members of the ARDRN strongly recommend these community-set research priorities are leveraged by the MRFF Missions Program to inform funding decisions moving forward and ensure public funding for research is directed towards what matters most to end users.

To RVA's knowledge and based on a survey of members of the Australian Rare Disease Research Network (ARDRN), funding for rare disease research through the MRFF Missions Program has come only from the Genomics Health Future Mission (GHFM). RVA acknowledges this may not be the whole picture; however, the MRFF does not routinely record whether a project is funding rare disease research, so there is currently no accurate way to measure how the missions program responds to rare disease. This leaves clear gaps in the other Missions Program responses to rare disease. For example, despite the significant mental health burden of rare disease, and the need for a nuanced response, to RVA's knowledge, based on sector feedback, the mental health mission program has not adequately responded to rare diseases.

The Impact of the MRFF Missions Program on Rare Disease Research

Programs such as **Mackenzie's Mission, UDN-Aus, acute care genomics, and genomic NBS** projects have delivered life-changing new diagnoses for families. However, the key issue for these programs is sustainability—once research funding ends, access and impact often end with it.

Policy alignment is critical to effective translation and equity. Stronger links between research and healthcare, supported clinician-researcher roles, and readiness of the policy environment are essential. Despite this, RVA has witnessed high-quality evidence for low cost, life changing interventions, borne out of Missions Program funding, fail to translate because of gaps in policy.

Ineffective Research Translation in Rare Disease Perpetuates Existing Inequities

Acute care genomics is one example of research, funded through the GHFM, that generated irrefutable evidence for a cost-effective lifesaving diagnostic pathway yielding substantial health system benefits. Tragically, a lack of policy readiness at the end of the research led to inequities in implementation across Australia because there are systemic policy gaps preventing nationally coordinated health system funding for implementation. That is why the MRFF should work towards ensuring new knowledge generated from the Missions Program does perpetuate systemic inequities.

In line with Recommendation two in this submission, the MRFF should require assessments of policy readiness to ensure policy environments are ready for equitable implementation of new knowledge. This would mean researchers are not left scrambling and speaking to policy experts when inequity has already taken hold.

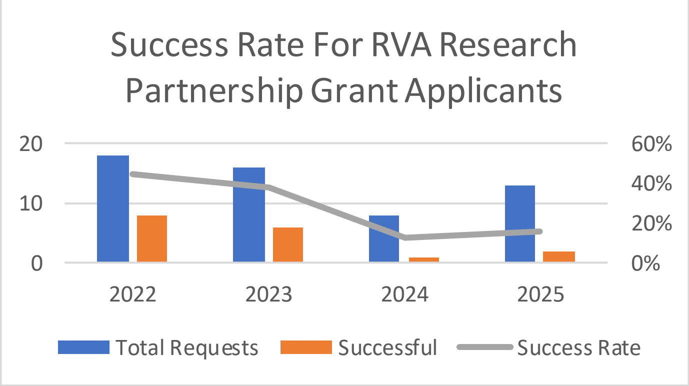
Acknowledging that some research may not uncover all the evidence required for effective translation, as a translation focused fund, the MRFF Missions Program should look for ways to ensure evidence from funded projects informs policy gaps.

RVA's Role and Broad Sector Knowledge of Rare Disease Research Funded Through the Missions Program

Though the RVA Research partnerships program RVA has been involved in MRFF Missions Program-funded research in advisory, co-design, and governance roles both through budgeted and in-kind contributions. RVA provides rare disease policy expertise and broad rare disease consumer expertise to research and acts as a conduit to the broader rare disease community. RVA also holds Associate Investigator and consortium advisory roles, including through GHFM targeted calls.

Interestingly, in 2023 through the RVA Research Partnerships program, RVA recorded an uptick in successful rare disease research applications following the targeted call for genomic newborn screening research via the GHFM. This is a testament to the importance of extending the Missions Program for rare disease research, a view shared by members of the ARDRN. However, since then, RVA has noted a concerning decline in the number of successful rare disease research applications across 2024 and 2025, via the RVA Research Partnerships program (Figure 1).

Figure 1. Success Rates for RVA Research Partnership Grant Applications (2022 –2025)



At present, based on RVA's peak body knowledge of the sector, despite public funding initiatives and streams targeting rare diseases, very few rare disease focused grants, with the exception of rare cancers, are successful. This may signal an issue at the review stage. It is possible reviewers assume, based on the name, the impact of these rare disease research will be minimal due to small numbers and therefore not worth the return on investment. This is a dangerous misconception, if we look at the estimated burden of rare diseases mentioned above. Indeed, solutions for rare diseases are often scalable & transferable across the health system. Investment in RD research can drive innovation with benefits for common conditions as well. Research addressing more common health challenges is not inherently more impactful than research targeting RD, where the stakes for individuals & families are often profound.

The Impact of the Missions Program According to the Australian Rare Disease Research Network

In December 2025, RVA surveyed members of the Australian Rare Disease Research Network (ARDRN) to gather their collective experience with the MRFF Missions Program. Fifteen ARDRN members responded to the survey. Of these, 10 ARDRN members mentioned they had received, or were receiving, funding for their research through the Missions Program, exclusively via the GHFM. All respondents want to see the Missions Program continue. Respondents consistently reported that Missions Program funding has been a significant enabler of collaboration across the Australian rare disease research ecosystem.

Overall, the Missions Program was seen as instrumental in fostering collaboration, national coordination and cross-project and cross-disciplinary networks within genomics, including initiatives such as RD-Now, RNA4RD, RD-Mass Spec, and Shariant, strengthening Australia's rare disease research capacity and impact.

Funding also supported the development of shared national infrastructure, such as CaRDinal, which is now used by more than 38 research studies and nearly 300 researchers and clinicians. This has strengthened coordination, standardised analysis and data management approaches, and enabled discoveries, including new disease genes, which would have been unlikely through isolated projects.

The funding also facilitated meaningful connections across multiple MRFF-funded rare disease projects, fostering collaboration across functional genomics, translation, and diverse rare disease groups. Importantly, respondents highlighted expanded engagement with conditions not routinely investigated for genetic causes, such as cerebral palsy, broadening the scope and inclusivity of rare disease research.

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