Implementing the National Strategic Action Plan for Rare Diseases

May 2023 Status Report

Acknowledgement of Country
We acknowledge Aboriginal and Torres Strait Islander peoples as the Traditional Owners of Country throughout Australia and their continuing connection to both their land and seas. We also pay our respects to Elders – past and present – and generations of Aboriginal and Torres Strait Islander peoples now and into the future.

Contents

About Rare Voices Australia

Rare Voices Australia (RVA) is the national peak body for Australians living with a rare disease. RVA provides a strong, unified voice to advocate for policy as well as health, disability and other systems that work for people living with a rare disease. RVA's work is non-disease specific and is based on the commonalities of rare disease. RVA is dedicated to working with all key stakeholders to drive the best outcomes for all Australians living with a rare disease through collaborative leadership for the development and implementation of rare disease policy in Australia. Key stakeholders include people living with a rare disease, governments, key peak bodies, researchers, clinicians and industry. In 2018, the Australian Government commissioned RVA to lead the collaborative development of the National Strategic Action Plan for Rare Diseases (the Action Plan).[1]

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, as well as other stakeholders, in leading the collaborative implementation of the Action Plan.

www.rarevoices.org.au

National Strategic Action Plan for Rare Diseases

Launched in 2020, the Action Plan's vision is to achieve the best possible health and wellbeing outcomes for the estimated 2 million Australians living with a rare disease. The Action Plan provides guidance and direction centred around three core Pillars – 1) Awareness and Education, 2) Care and Support, and 3) Research and Data. Priorities, actions and implementation areas are outlined under each pillar for consideration by a wide range of stakeholders, including governments at all levels, non-government organisations, the public and private health sectors, industry, researchers and academics, rare disease organisations and the wider community. Implementation of the Action Plan is the responsibility of all stakeholders, with the uptake of the outlined recommendations being an individual decision for each stakeholder, based upon their area of responsibility, governance remit, existing activities and future planning and directions in relation to rare diseases. The sector has embraced this responsibility and implementation is underway.

Authors (The authors contributed equally to this work)

Dr Falak Helwani

Falak is the Research and Evaluation Manager at RVA. She is a former molecular cell biologist with lived experience as a parent to two children living with a in rare disease. Falak has a PhD in cell biology from the Institute for Molecular Bioscience at the University of Queensland and has co-authored several peer-reviewed papers. She also has 6 years of postdoctoral experience in stem cell research at the Mater Medical Research Institute in Queensland. In her role at RVA, Falak is involved in the development of the RARE Portal, management of RVA research partnerships and has co-authored a White Paper and Strategy for Australia's Rare Metabolic Workforce. She is also involved in several research projects progressing different areas of the Action Plan.

Dr Amanda Choo

Amanda is the Resources and Information Officer at RVA. She has been involved in scientific research since 2008 and has worked on projects involving rare diseases. Amanda has a PhD in the area of molecular genetics from the University of Adelaide as well as 7 years of postdoctoral experience in genetic research. In her role at RVA, Amanda is involved in the development of the RARE Portal. She also has lived experience in rare disease.

EXECUTIVE SUMMARY

The Australian Government's National Strategic Action Plan for Rare Diseases (the Action Plan), launched in 2020, is the first nationally coordinated effort to address rare diseases in Australia.[1] Implementation of the Action Plan is the shared responsibility of the entire rare disease sector, including people living with a rare disease, governments, key peak bodies, clinicians, researchers and industry. As the national peak body for Australians living with a rare disease, Rare Voices Australia (RVA) is leading the collaborative implementation of the Action Plan.

Monitoring implementation of the Action Plan

Between September and October 2022 and during February 2023, RVA conducted an activity scan—the first measure of Action Plan progress since it was launched in 2020—inviting the rare disease sector to share their projects, initiatives and achievements.

The activity scan captured hundreds of activities aligning with Action Plan Pillars and Priorities. These activities were categorised according to 5 key elements of progress—Input, Activities or Processes, Outputs, Outcomes and Impact. The activities were also mapped to the 8 key themes in the Action Plan, which were developed, by the rare disease sector, for the rare disease sector, as descriptive indicators of progress.

Key findings

Sector-wide activity scan: Setting the baseline, identifying gaps and highlighting strengths

- Rare disease sector activities strongly align with Action Plan Pillars, Priorities, Actions and Themes, demonstrating that the Action Plan has mobilised the sector to work on targeted/common goals
- Some activities are being translated into output and outcomes; this is anticipated to increase in future
- At this stage, impact is difficult to measure and can be subjective. Person-centred, verified indicators or metrics are needed to facilitate objective measurements of impact in rare disease. One of the key ways to achieve impact is systemic adoption of evidence-based strategies through policy reform.
- In identifying areas with more significant gaps in Action Plan implementation, fewer activities were captured across the following 4 Themes compared to other Themes:
    - Theme 4: The need to measure rare diseases;
    - Theme 5: The need for sustainable systems and workforce;
    - Theme 7: State, national and international partnerships as well as cross sector (i.e. specifically across government sectors) collaboration; and
    - Theme 8: The need to progress early implementation wherever possible.

Recommendations

All governments must urgently invest in the rare disease sector. This should include investment in regular reviews of Action Plan progress to support iterative implementation plans for a responsive, dynamic, transformative and targeted approach.

The sector must be supported to collectively and continually:

- prioritise gaps;
- ensure activities address priorities across more than one Action Plan Pillar;
- translate inputs, activities and outputs into outcomes and impact;

- work towards systemic change and prioritisation of broader impact;
- count rare diseases in Australia;
- progress sustainable systems and workforce for all rare diseases;
- ensure cross-system collaboration and partnerships;
- facilitate early implementation;
- address the specific needs of priority populations identified in the Action Plan; and
- foster a culture of evaluation.

ABOUT THE RESEARCH

Objectives

1. Track implementation of the Action Plan since its launch in 2020.
2. Identify projects, initiatives and achievements of various stakeholders and, where relevant, align these against the Pillars, Priorities and Themes in the Action Plan and map these to 5 key elements of progress in a logic model (See Figure 1).[2]
3. Identify gaps and strengths in Action Plan implementation to guide the sector towards the more effective and efficient use of time, expertise and resources.
4. Set a baseline for future monitoring and evaluation of Action Plan progress.

Methods

Data collection

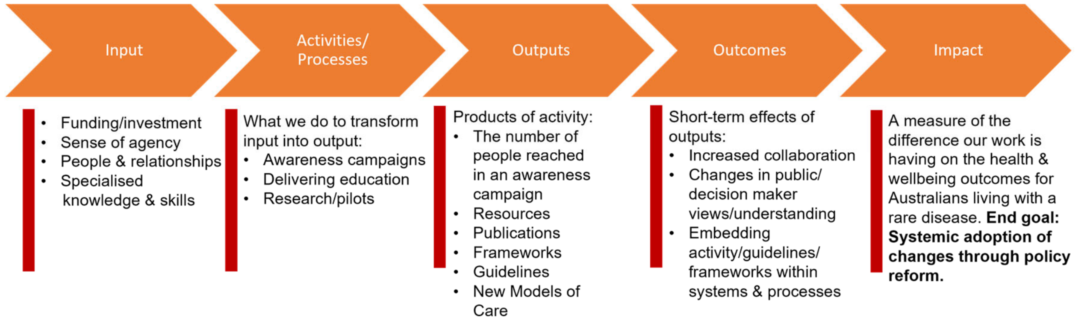
Between September and October 2022 and during February 2023, RVA invited rare disease stakeholders—departments of health, clinicians, researchers, industry, key peak bodies and RVA Partner organisations (rare disease groups)— to share work undertaken since the Action Plan's launch in 2020. Contributions included projects, initiatives, events, completed resources, funding schemes, policies, publications and other achievements. For ease of discussion throughout this document, these are collectively referred to as 'activities'. Contributions were made via Google Jamboard (a digital interactive whiteboard) or an online submission form on RVA's website. Email contributions were also accepted.

Data analysis

Individual activities were assigned to the 3 core Pillars of the Action Plan[1] – Pillar 1: Awareness and Education, Pillar 2: Care and Support, and Pillar 3: Research and Data. They were further categorised across specific priorities under each of these 3 Pillars, with some activities aligning to more than one Priority and/or Pillar. See Appendix B for a summary of Action Plan Pillars and Priorities.

Activities were then grouped under 5 key elements of progress—Input, Activities or Processes, Outputs, Outcomes and Impact (Figure 1). The number of activities under each key element of progress was quantified for each Pillar. Activities were also aligned against the 8 key themes in the Action Plan, which were developed by the rare disease sector, for the rare disease sector as descriptive indicators of Action Plan implementation progress (Table 1). To understand implementation progress at the level of the Action Plan Pillars, the number of activities aligning with each key theme under each Pillar was also quantified. Preliminary findings were presented at the 2022 National Rare Disease Summit (the Summit). Findings in this report are an extension of those shared at the Summit.

Figure 1. Logic Model: Defining Elements of Progress



- Funding/investment
- Sense of agency
- People & relationships
- Specialised knowledge & skills

What we do to transform input into output:
- Awareness campaigns
- Delivering education
- Research/pilots

Products of activity:
- The number of people reached in an awareness campaign
- Resources
- Publications
- Frameworks
- Guidelines
- New Models of Care

Short-term effects of outputs:
- Increased collaboration
- Changes in public/ decision maker views/understanding
- Embedding activity/guidelines/ frameworks within systems & processes

A measure of the difference our work is having on the health & wellbeing outcomes for Australians living with a rare disease. **End goal: Systemic adoption of changes through policy reform.**

Note: This logic model consists of 5 key elements of progress[2] —Inputs, Activities and Processes, Outputs, Outcomes and Impact—where the greatest impact is attained through systemic reform and the embodiment of the Action Plan's vision.

Table 1. Action Plan Themes

Action Plan Themes
Theme #1: The need for national leadership, coordination and consistency
Theme #2: The need to prioritise the systematic building of knowledge, evidence and expertise
Theme #3: The need for a person-centred approach and ongoing collaboration
Theme #4: The need to measure rare diseases
Theme #5: The need for sustainable systems and workforce
Theme #6: The need for stakeholder collaboration
Theme #7: State, national and international partnerships as well as cross-sector *(i.e. specifically, across government sectors)* collaboration
Theme #8: The need to progress early implementation wherever possible

Note. Action Plan Themes are descriptive indicators of progress informed by consultation with the rare disease sector.[1]

INTRODUCTION

The launch of the National Strategic Action Plan for Rare Diseases (the Action Plan) in 2020 was a significant milestone for Australians living with a rare disease. The Australian Government's Action Plan is the first nationally coordinated effort to address rare diseases in Australia. Before the Action Plan, progress in the rare disease sector was largely fragmented and siloed.

Collaborative development of the Action Plan, led by RVA, highlighted several issues, including lack of awareness about rare diseases, the struggle for a timely and accurate diagnosis, limited care and support options, a lack of research, as well as poor data collection and use.

The Action Plan's detailed person-centred pillars, priorities, actions and implementations steps have strategically steered the sector's activities. The value in leveraging the Action Plan to guide and unite the sector has been echoed by all stakeholders. The Action Plan is increasingly mobilising the sector to work collectively towards its common vision – '*The best possible health and wellbeing outcomes for people living with a rare disease*'. Cross-sector collaboration is vital for true reform and effective impact in achieving this vision.

Between September and October 2022 and during February 2023, RVA led a sector-wide activity scan in the first effort to track implementation of the Action Plan. The aim of this activity scan was to start mapping what is being done, what has been achieved, and importantly, to highlight gaps and opportunities to maintain momentum, prevent duplication and encourage collaboration.

Over 380 activities were captured during this initial scan, all aligning with different Pillars, Priorities and Themes in the Action Plan. The activities were collated and analysed in various ways to identify gaps and highlight sector achievements. This report provides a detailed account of the activity scan methodology, results and next steps to progressing Action Plan implementation.

RESULTS

Mapping rare disease sector activities to Action Plan Pillars and Priorities

The activity scan captured over 380 activities from across the rare disease sector aligning with one or more Pillars and Priorities in the Action Plan. Some activities broadly addressed all rare diseases, while others related to a single rare disease, or an umbrella group of rare diseases. Activities were either at a national or jurisdictional level. The vast majority of activities were those undertaken by RVA Partner organisations (individual rare disease organisations). This is not surprising as this stakeholder group is the largest of those invited to contribute to this research, which included departments of health, clinicians and researchers, industry and peak bodies.

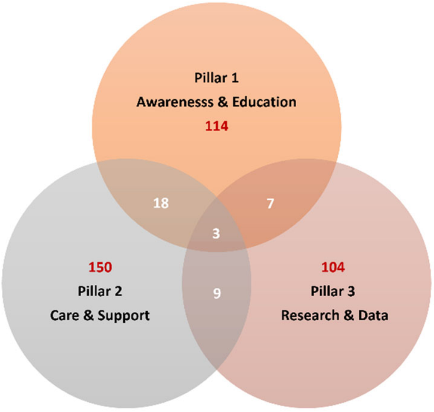
Action Plan Pillars are easily recognisable to people living with a rare disease, and whilst each of these is considered separately in the Action Plan, in reality, all Pillars are interrelated. As such, the strongest policy responses address priorities across multiple Pillars. Effective policy reform in one area will create change and momentum in other areas. Accordingly, activities that respond to multiple priorities and pillars should be prioritised.

Figure 2 shows the number of captured activities aligning with each Action Plan Pillar. Only a fraction of these activities address multiple Pillars (and priorities) simultaneously. More activities respond to both Pillar 1: Awareness and Education and Pillar 2: Care and Support compared with activities overlapping between these Pillars and Pillar 3: Research and Data. Figure 3 provides a more granular look at the number of activities addressing each individual Priority underneath these Pillars.

With most activities addressing Pillar 2: Care and Support and the majority being conducted by individual rare disease organisations, it is apparent that these organisations are taking on a number of care and support roles across the rare disease sector. In line with this observation, through RVA's mentorship and education program, RVA is aware of the significant burden on many rare disease organisations as they work to fill gaps in the care and support needs of patients and their families.

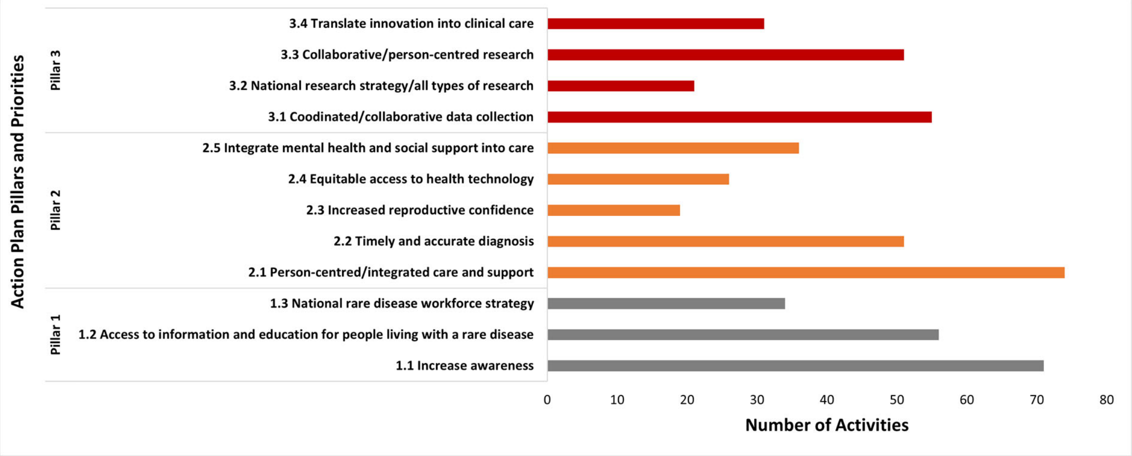
Taken together, these data highlight the role of the Action Plan as a framework driving hundreds of activities across the sector. The Action Plan has mobilised the sector to work on shared goals, proving the power of a plan, developed by the rare disease sector, for the rare disease sector, that responds to common gaps and challenges in rare disease.

Figure 2. Alignment and Overlap of Activities with Action Plan Pillars



Note: Venn diagram showing how the captured activities align with the three Action Plan Pillars. The red numbers denote the total number of activities addressing a single Pillar. The white numbers denote the number of activities that traverse the Pillars.

Figure 3. Alignment of Activities with Action Plan Priorities



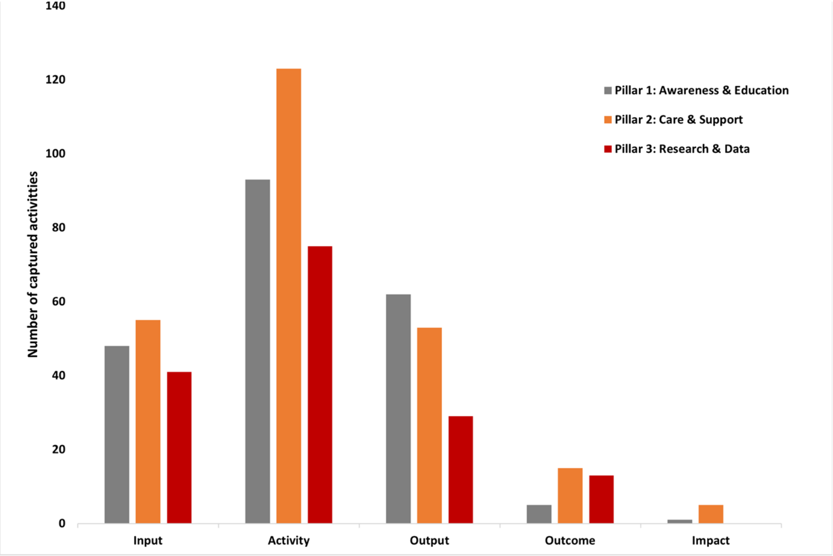
Note: Bar graph showing how captured activities align with the priorities across Action Plan Pillars. The grey bars represent activities aligning to Pillar 1, the orange bars represent activities aligning to Pillar 2 and the red bars represent activities aligning to Pillar 3.

Tracking activities against 5 key elements of progress

The vision to achieve ‘the best possible health and wellbeing outcomes for Australians living with a rare disease’ is the impetus and aspiration behind all work progressing the Action Plan. This vision can only be realised through stages of implementation. To evaluate sector progress in achieving the Action Plan’s vision, activities captured were mapped to 5 key elements of progress in a logic model—Inputs, Activities and Processes, Outputs, Outcomes and Impact—where the greatest Impact is attained through systemic reform and the embodiment of the Action Plan’s vision (Figure 1).

There is a consistent trend in the alignment of captured activities to the 5 key elements of progress across all three Action Plan Pillars (Figure 4), with most—regardless of the Pillar—in the Activity and Output stages and fewer in the later stages of Outcome and Impact. This points to similarities in sector progress in all areas of the Action Plan. Collectively, close to half the captured activities are in the ‘Activities or Process’ phase of the logic model, while only a fraction (approximately 5%) have reached the Impact phase (Figure 4). This is not unexpected 3 years into Action Plan implementation. Given the natural progression of the logic model continuum from Input through to Impact, the sector is likely to see many of the current Activities and Processes translate to Outputs, Outcomes and measurable Impact in the near future. This will require increased levels of collaboration and a culture of evaluation across the sector to ensure all work leads to meaningful and measurable impact for the best possible health and wellbeing outcomes for Australians living with a rare disease.

Figure 4. Alignment of Activities with the 5 key elements of progress



Note: Bar graph showing the number of activities according to each Action Plan Pillar that align to the elements of progress—Inputs, Activities and Processes, Outputs, Outcomes and Impact. The grey bars represent activities in Pillar 1, orange for Pillar 2 and red for Pillar 3.

Tracking activities against Action Plan Themes

Collaborative development of the Action Plan identified 8 themes to be descriptive measures of progress.[1] These themes are detailed in Table 1 of this report. Assignment of captured activities to the 8 themes was guided by a strict set of criteria (See Appendix A). The majority of activities align with more than one Action Plan Theme, which meets the shared expectations of the sector. The greatest levels of activity alignment are across the following themes:

- Theme 1: National leadership, coordination and consistency;
- Theme 2: Prioritisation of systematic building of knowledge, evidence and expertise;
- Theme 3: A person-centred approach; and
- Theme 6: Stakeholder collaboration.

By comparison**,** fewer activities align with:

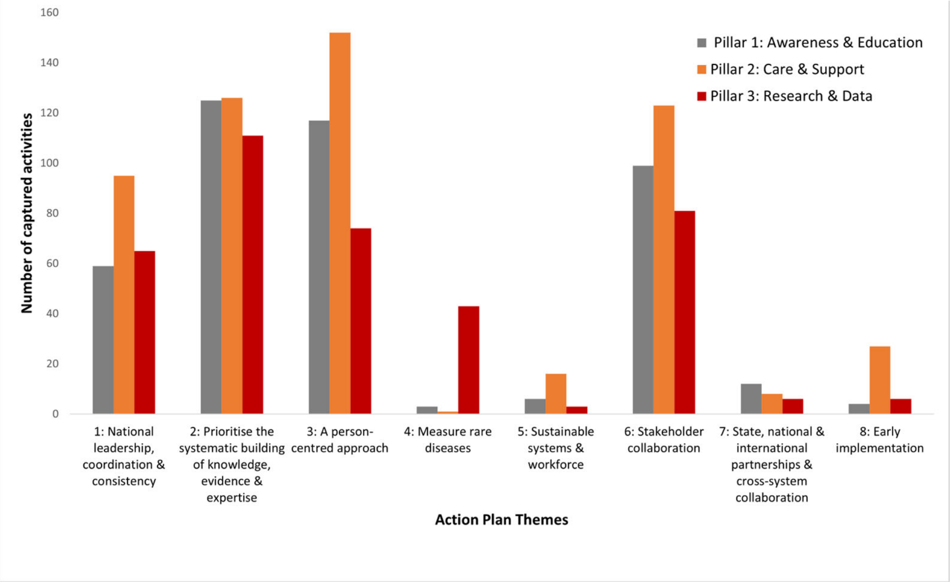
- Theme 4: Rare disease measurement;
- Theme 5: Sustainable systems and workforce;
- Theme 7: State, national and international cross-sector (i.e. specifically across government sectors) collaboration; and
- Theme 8: Progression of early implementation wherever possible.

These data also speak to the relationships between the Action Plan Themes and Pillars across all captured activities. Some Pillars are inherently aligned with particular themes; however, there are some themes requiring more attention in specific Pillars. Unsurprisingly, most activities aligning with Theme 4: 'Rare disease measurement' fall under Pillar 3: Research and Data. For Theme 5: 'Sustainable systems and workforce' and Theme 8: 'Progression of early implementation wherever possible', Pillar 2: Care and Support activities are most prominent, pointing to greater levels of progress in the activities under this Pillar compared to other Action Plan Pillars (Figure 6).

In the next phase of Action Plan implementation, it is important to prioritise the measurement of rare diseases through coordinated data collection, and the development of sustainable systems and workforce, which are identified across the Action Plan in the critical enablers, priorities and actions as well as the Action Plan Themes. The sector also needs to think of ways to work more collaboratively across government sectors, jurisdictions and internationally, and push for early implementation wherever possible. The need for 'rare disease care and support that is integrated, incorporating clear pathways throughout health, disability and other systems' (Action 2.1.1) is highlighted under Pillar 2: Care and Support and is becoming increasingly prioritised in some jurisdictions working to connect people living with a rare disease to all areas of government support. It is vital that this work is extended to all jurisdictions across the country.

Of note, fewer than 20 captured activities appear to address priority populations outlined in the more granular implementation steps of the Action Plan Action Plan. Future measurements of Action Plan implementation should include specific questions around how individual activities have considered the needs of these priority populations to allow stronger conclusions to be drawn about progress in these areas.

Figure 6. Alignment of Activities with Action Plan Themes



Note: Bar graph showing the number of activities aligning with the Themes under each Action Plan Pillar.

LIMITATIONS OF THE RESEARCH

This is an initial attempt to map the rare disease sector's progress implementing the Action Plan, and there are a few limitations that should be considered in any interpretations of the findings.

Sample bias

The results herein are based on a sample of sector-wide activities. This sample was dependent on RVA's reach through existing networks, the receipt of the initial email invitation (the email went into the Junk folders of some stakeholders), the knowledge, willingness and capacity of stakeholders to contribute, as well RVA's knowledge of activities across the sector. Various efforts were made to increase the sample size and scope. Stakeholders were invited via targeted emails, RVA's monthly e-newsletter and social media to contribute using 2 mediums—Google Jamboards (a digital interactive whiteboards) and direct email. In response to feedback from our largest stakeholder group—RVA Partner organisations—a second invitation was sent to contribute via a Gravity Form (online form). Establishing more effective ways to facilitate stakeholder participation is important to increase the sample size for future monitoring of Action Plan implementation.

The number of activities contributed by RVA Partner organisations outnumbered those of other stakeholder groups, introducing possible undercoverage bias for other stakeholder groups. This limitation is hard to avoid as the number of RVA Partner organisations (over 100 at the time of writing this status report), and naturally, the number of rare diseases will always outnumber other stakeholders and their countable activities. However, activities of governments, clinicians, researchers and other stakeholders are often applicable to a broad range of rare diseases. For example, the work of Mackenzie's Mission and many other pilot projects are built with intentions for broader impact. Developing ways to empirically measure the broad applicability of such activities should be considered in future measures of Action Plan progress.

Researchers' interpretations of activities

Data analysis was dependent on the clarity and extent of information provided or available about each activity. In cases where deep knowledge or exact details of activities were not shared by stakeholders, interpretation and assignment of activities to various Action Plan Pillars, Priorities and Themes became difficult and at risk of subjective bias. To overcome this and ensure consistency in interpreting and categorising activities, stringent criteria were developed (See Appendix A), and 2 researchers coded and cross-checked the results. Future interpretation of the impact of activities would be improved by extending the logic model to include 'reach', which will capture the stakeholder and jurisdictional scope of activities.[3]

Data collection methods

This first attempt to map the sector's progress implementing the Action Plan has highlighted the types of information that may be useful in understanding and analysing rare disease sector activities. Future activity scans will leverage these learnings to develop more targeted ways of gathering the information for streamlined data analysis and robust conclusions. One possible way forward is to build forms or a survey tool to guide stakeholders through the details of what to share as part of their contribution of activities. This tool should include questions related to the stakeholder group; the jurisdictional scope of activities, as a new indicator of reach; and also questions around how any activities support priority populations in the Action Plan (e.g. people living with an undiagnosed rare disease, Aboriginal and Torres Strait Islanders, members of the culturally and linguistically diverse community, and people living in rural and remote areas). These amendments will reduce subjective biases in the interpretation of activities and ensure a more accurate and complete snapshot of implementation progress.

External influences

This activity scan has not considered external factors that may influence implementation progress. There are likely to be several factors, not explored through this activity scan, that have either supported or hindered implementation. Whilst it is important to consider these factors, this was outside the scope of this research. Therefore, based on the results herein, no comment can be made on the reasons for different levels of progress across different areas. Moving forward, any enablers and barriers to Action Plan implementation should be carefully considered by all stakeholders in response to the basic findings presented herein. Leveraging enablers and actively addressing barriers will support implementation progress.

Observer bias

The final key element of progress, 'Impact', is challenging to measure as impact may be perceived differently by different stakeholders. To manage observer biases in categorising activities as having made an impact and across all 5 key elements and 8 Themes, criteria were developed to ensure consistency (See Appendix A), and 2 researchers collected, processed, and analysed the data separately and cross-checked results—this is known as investigator triangulation.[4] A set of rare disease specific, validated indicators would be useful for evaluating impact and managing biases; however, these remain to be developed.

SUMMARY

Between October 2022 and February 2023, as the national peak body for Australians living with a rare disease leading the collaborative implementation of the Action Plan, RVA conducted an activity scan. The aim was to track collective progress of the rare disease sector in implementing the Action Plan since its launch in 2020. This activity scan provides a snapshot of what is being done, what has been achieved, and importantly, it highlights relative gaps in progress.

The great complexity and unmet need in rare diseases can be overwhelming for the entire sector, including policymakers, clinicians, practitioners, researchers, academics, industry and especially for the people who live with a rare disease. The Australian Government's' Action Plan, the first nationally coordinated effort to address rare diseases in Australia, was developed to address this unmet need and reduce uncertainly through policy.

The power of an Action Plan developed by the rare disease sector for the rare disease sector was expressed by a broad range of stakeholders at the 2022 National Rare Disease Summit[5] (the Summit) who champion the Action Plan as a common ground or anchor for their work. Stakeholders at the Summit said:

> "There is legitimacy given to rare disease support groups who are now seen and heard through the Action Plan and RVA's work to unify the voices of people living with a rare disease." – **patient advocate**
>
> "The Action Plan is a framework and useful tool for clinicians and the wider healthcare sector to increase awareness of rare diseases, respond to gaps and advocate for the needs of patients." – **clinician**
>
> "The Action Plan has encouraged more patient-centred and collaborative research and raised awareness among government funding bodies. It has highlighted the need for research to address both commonalities and nuances of rare diseases." – **medical researcher**
>
> "The Action Plan has helped inform approaches to rare diseases and more widely anchors the actions and decisions of industry."[5]– **industry representative**

Using the activity scan to track Action Plan implementation progress, RVA has had the opportunity to pause and reflect on the tireless efforts of stakeholders across the rare disease sector. In 3 years, and through the challenges of a global pandemic, the accomplishments of the sector are commendable. What is clear from the results is that activity relates to need – more immediate needs engender a higher degree of activity—as seen in the higher level of activities aligned with Action Plan Pillar 2: Care and Support. However, there are several gaps in Action Plan implementation and opportunities to learn and improve. Specific areas currently lacking—as evidenced by the smaller numbers of activities aligning by comparison—include Pillar 3: Research and Data, the measurement of rare diseases (Theme 4), sustainable systems and workforce (Theme 5), cross-sector collaboration specific to different government sectors (Theme 7), and the progression of early implementation activities (Theme 8). Moving forward, these gaps must be addressed to ensure full implementation of the Action Plan and equitable opportunity across the rare disease sector. Increased collaboration across different stakeholders and within stakeholder groups should also be prioritised to avoid duplication of activities and encourage the efficient use of resources.

This status report of Action Plan progress is a means of introspection for the whole sector. The results should support all stakeholders to review and refine their approaches for the most effective and efficient path to early implementation of sustainable and systemic change across all Action Plan pillars, priorities, actions and implementation steps.

Responding to the Action Plan is not a linear process, but rather a continuous cycle of monitoring, evaluation, learning and refinement. Fostering a culture of evaluation across the rare disease sector will ensure all activities remain aligned with the Action Plan. This is vital to perpetual learning across the sector and to refining approaches for more meaningful impact towards achieving the vision of the Action Plan - '*The best possible health and wellbeing outcomes for people living with a rare disease*'.

Maintaining momentum: Things to consider for sustainable and systemic change

This initial, more comprehensive, approach to mapping progress has enabled clarification of RVA's methods for measuring Action Plan implementation progress. It has led to the establishment of a baseline for more streamlined and targeted monitoring, evaluation and reporting on implementation progress. This baseline will be used in future evaluations to track implementation trajectory over time. However, it should be acknowledged that without verified indicators or metrics to assess and track progress across the rare disease sector, measuring impact can be subjective, particularly given the varied connotations of impact at an individual level compared to a sector-wide level. Person-centred, verified indicators or metrics are needed to facilitate objective measurements of impact in rare disease.

Future efforts to monitor and evaluate Action Plan progress should be refined to increase the sample of captured activities through improved data collection methods. In addition, data analysis and reporting should be streamlined to enable the sharing of simple progress snapshots over time. This work requires ongoing commitment of the rare disease sector to track Action Plan progress, and to iterative monitoring, evaluation and refinement of individual activities to meet intended outcomes.

Moving forward, it is important to think about whether the sector should be doing more, doing better, or both, in progressing Action Plan implementation. Based on the findings of this activity scan, the sector must collectively and continually review the following questions.

How does the sector:

- prioritise gaps?
- ensure activities address priorities across more than one Pillar?
- translate input, activities and outputs into outcome and impact?
- work towards systemic change and prioritisation of broader impact?
- count rare diseases in Australia?
- progress sustainable systems and workforce for all rare diseases?
- ensure cross-system collaboration and partnerships?
- facilitate early implementation?
- address the specific needs of the priority populations outlined in the Action Plan?

All governments must urgently invest in the rare disease sector. To drive systemic change, investments should leverage and build on existing expertise, knowledge, resources and infrastructure. Learnings from this activity scan also highlight the need to invest in regular reviews of

Action Plan progress to support iterative implementation plans for a responsive, dynamic, transformative and targeted approach.

Implementation of the Action Plan is the ongoing responsibility of all stakeholders, including all levels of government, the public and private health sectors, rare disease organisations, industry, researchers and the wider community. RVA remains committed to leading the collaborative implementation of the Action Plan and will continue to monitor and evaluate progress and steer the sector to the realisation of the collective vision for the best possible health and wellbeing outcomes for Australians living with a rare disease.

REFERENCES

1. Commonwealth of Australia. Department of Health. National Strategic Action Plan for Rare Diseases. Canberra; 2020. 63 p. Available from: https://www.health.gov.au/sites/default/files/documents/2020/03/ national-strategic-action-plan-for-rare-diseases.pdf
2. New South Wales Government. NSW Ministry of Health, Centre for Epidemiology and Evidence. Developing and Using Program Logic: A Guide. Evidence and Evaluation Guidance Series, Population and Public Health Division. Sydney; 2017. 18 p. Available from: https://www.health.nsw.gov.au/research/Publications/developing-program-logic.pdf
3. Montague S, Porteous NL. The case for including reach as a key element of program theory. Eval. Program Plann. [Internet]. 2013;33(1):177–83. Available from: https://doi.org/10.1016/j.evalprogplan.2012.03.005
4. Carter N, Bryant-Lukosius D, DiCenso A, Blythe J, Neville AJ. The use of triangulation in qualitative research. Oncol. Nurs. Forum [Internet]. 2014;41(5):545–47. Available from: http://doi.org/10.1188/14.onf.545-547.
5. Rare Voices Australia. Summary of the 2022 National Rare Disease Summit. 2022. 10 p. Available from: https://rarevoices.org.au/wp-content/uploads/2022/12/2022NationalRDSummitSummary.pdf

APPENDICES

Appendix A: Rationale for tracking activities against different indicators of progress

Rational for Coding Activities Against 5 Key Elements of Action Plan Progress – Logic model

Elements of Progress	Types of Activities
Input	Examples include funding and investment from various sources, sense of agency, people and relationships, the contribution of time, specialised knowledge and skills.
Activities/Processes	What we do to transform input into output. For example, awareness campaigns, delivering education, research, pilot projects.
Outputs	Products of activity. For example, the number of people reached in an awareness campaign, resources, publications, frameworks, guidelines, new Models of Care.
Outcomes	Implementation of outputs. Short-term effects of outputs, including increased collaboration, changes in public/decision maker views and/or understanding. Embedding activity/guidelines/frameworks within systems and processes including care standards.
Impact	A measure of the difference our work is having on the health and wellbeing outcomes for Australians living with a rare disease. One example of an end goal is the systemic adoption of changes through policy reform.

Rationale for Coding Sector Progress Against the 8 Themes Mentioned in the Action Plan as Measures of Progress

Action Plan Themes	Theme Descriptions	Types of Activities Aligning with This Theme
Theme #1: The need for national leadership, coordination and consistency	This theme emerged most frequently from the consultation process, with participants calling for: • A **national plan for Australia** that is in line with global standards. Australia remains in danger of falling further behind many countries already tackling rare diseases through policy and legislation. This Action Plan is an opportunity for Australia to adopt a national plan for rare diseases that aligns with global standards. The value of a nationally coordinated plan cannot be underestimated. • **Annual implementation plan** Existing plans for rare diseases, including those in Europe and the United Kingdom (UK), are accompanied by an implementation plan. In Australia, this plan should be developed collaboratively by the sector and could be led by the national peak body for Australians living with a rare disease. Progress in implementation should be regularly monitored and reviewed. • **Ongoing stewardship and policy sustainability** Ongoing stewardship of the Action Plan is critical to ensure policy change is long-lasting and sustainable. In the United States, UK and many European countries, the sustainability of rare diseases policy is enshrined **in legislation.**	• Forums of clinical leadership/expertise, • Government grants/mass funding/budget allocations (Genomic Health Future Mission rounds) • Funding for any research projects • Nationally led resources (e.g. Department of Health resources) • Centres of Excellence/Expertise • Any activities associated with a Parliamentary Inquiry • Any activities that involve engagement with government? • MoC/Clinical care guidelines • MBS/PBS funded items • Peak body work, including all RVA's activities as a national peak body (i.e. RARE Portal, Education Portal, RArEST project etc) • National strategies • National screening programs • National leadership for priority populations • National frameworks and policies • UN resolutions

Action Plan Themes	Theme Descriptions	Types of Activities Aligning with This Theme
		• Nationwide awareness activities, including national awareness days
Theme #2: The need to prioritise the systematic building of knowledge, evidence and expertise	There is urgent need for **the expansion of rare disease expertise** and **further development of evidence-based rare disease care**. **Systems** must actively respond to existing evidence gaps. **Processes that will build knowledge and evidence both quickly and sustainably** must be prioritised. **Clearer pathways through health and other systems** are a necessity. Throughout the consultation process, stakeholders consistently raised the need **for centres of excellence for rare diseases**. Currently, rare disease clinics and research institutes with a focus on rare diseases are significantly under-resourced, and often work in isolation. To achieve real progress, existing strengths must be built upon to formalise a network of centres of excellence that is appropriate and accessible for all Australians.	• All education activities/ materials/ resources/factsheets • Awareness days/activities • Centres of excellence/expertise • Forums and webinars • RVA Online Education Portal • Publications • Clinician education • Research • Submissions • Parliamentary Inquiry activities • Clinical care guidelines • RArEST activities • Peak body work • Registries • Active participation and input in conferences

Action Plan Themes	Theme Descriptions	Types of Activities Aligning with This Theme
Theme #3: The need for a person-centred approach and ongoing collaboration	To be successful, this Action Plan must progress meaningful involvement of people living with a rare disease across all areas. This includes **ongoing collaboration and co-design with the many rare disease organisations** that represent Australians living with a rare disease. These organisations enable connection and support, lead advocacy and awareness, and encourage active consumer participation. This Action Plan presents an important opportunity to embed the **rare disease consumer voice in the design, implementation and evaluation of services for Australians** at all levels. This has the potential to lead to better outcomes for people living with a rare disease as well as their families and carers	• Awareness days • Patient carer support and connection opportunities • Collaborative (co-designed) rare disease organisation resources • Inclusion of patient voice in different activities (e.g. Parliamentary Inquiry, health systems planning, resource development, clinical care guidelines/MoC, conferences) • Parliamentary Friends of Rare Diseases • Undiagnosed programs • Resources/support tailored for priority populations • Peak body work
Theme #4: The need to measure rare diseases	Limited data is a common feature in rare diseases. This is heightened by poor quality, disjointed collection methods and the ineffective use of data for rare diseases. Such limitations are evident across a range of areas, from health system classification to research. **Research, monitoring and ongoing evaluation** are critical in rare diseases because, ultimately, if we are not counting rare diseases, people living with rare diseases do not count	• Rare disease registries and databases • Policy or frameworks related to registries • Rare disease data coding efforts

Action Plan Themes	Theme Descriptions	Types of Activities Aligning with This Theme
Theme #5: The need for sustainable systems and workforce (defined as all types of workforce required for individuals living with rare disease, not only healthcare workforce)	**Sustainable systems and workforce** are critical to the long-term success of this Action Plan. Throughout stakeholder consultations, we heard many reports of staff shortages and a lack of funding. As such, there is a real need to **build on, and invest in, the existing strengths of the workforce for rare diseases**. **National leadership** is required to coordinate stakeholders to develop and implement a **workforce strategy** for rare diseases. The essential role of rare disease organisations must also be recognised and sustained. Rare disease organisations play a key role in raising disease awareness and providing much-needed person-centred information. These organisations often fill gaps in the system, not only in terms of awareness and education, but also in peer support and, increasingly, in the research and data sphere. However, these organisations are significantly under-resourced and are largely volunteer-based, posing a risk to their long-term sustainability.	• Metabolic Workforce White Paper/Strategy Pilot and peak body advocacy • Initiatives that highlight the need for sustainable systems and workforce, such as Rare Care Centre Pilot • Embedding rare disease expertise in the NDIS • Initiatives that ensure better care coordination, transition service and timely access to care/diagnosis • Any recognition of and sustainable funding for rare disease organisations/rare diseases registries and peak bodies • Any strategy for sustainable systems and workforce across the rare disease sector
Theme #6: The need for stakeholder collaboration (multi-stakeholder)	The success of this Action Plan is underpinned by **stakeholder involvement, collaboration and engagement**. It is essential that all key stakeholders in the rare disease community, including **people living with a rare disease, clinicians, researchers, governments and industry work together** to progress this Action Plan	• Awareness days • Research teams/collaborative projects • Multidisciplinary teams • Communities of Practice (e.g. RArEST activities, project ECHO) • RVA Education Portal • Submissions/consultations

Action Plan Themes	Theme Descriptions	Types of Activities Aligning with This Theme
		- Multistakeholder involvement in parliamentary inquiries/events - Parliamentary Friends of Rare Diseases - Any co-design projects (e.g. research, clinical care guidelines, strategies, MoC) - Alliances - Peak body work - Conferences that involve multi-stakeholder engagement
Theme #7: State, national and international partnerships as well as cross-sector [specifically across government sectors] collaboration*	Given the small populations and complexity involved in rare diseases, strong ongoing partnerships are invaluable**. Global collaboration and the sharing of knowledge and expertise** are often required to ensure the best outcomes for people living with a rare disease. Due to the nature of Australia's health and social systems, **state and national partnerships are vital, as is the need for the ongoing facilitation of these partnerships**. The complex nature of rare diseases requires **the integration of numerous public domains that extend beyond health to disability, social/welfare, education, employment, housing** and many other areas. At a national level, **policy leadership** is required to enable the effective and efficient delivery of integrated whole-of-life care that supports and responds to people's needs. Similarly, national policy	- Parliamentary Friends of Rare Diseases - Peak body work, including RVA advocacy - Any co-design rare disease projects/guidelines across public domains (e.g. Rare Care) Submissions/consultations and other activities that bring together public domains

Action Plan Themes	Theme Descriptions	Types of Activities Aligning with This Theme
	leadership is also required to seamlessly address health and social system challenges	
Theme #8: The need to progress early implementation wherever possible**	Rare diseases are often progressive and shorten life expectancy, and the burden of rare diseases remains unacceptably high. As such, **implementation activities must build on successful initiatives** already underway to address the need for urgency and to continue to build capacity, collaboration and coordination	• MBS/PBS funding e.g. for new treatments • National uptake of new diseases on the NBS • Timely national access to screening programs (e.g. Mackenzie's mission) • Mitochondrial Donation Law Reform (Maeve's Law) Act 2022 • Timely access to new health technologies – including via clinical trials • Research involving implementation of early intervention • Timely uptake of evidence-based strategies and recommendations (e.g. metabolic workforce strategy/national recommendations for rare disease registries) • Consultations to improve develop initiatives/frameworks/guidance

Abbreviations: Extension for Community Healthcare Outcomes (ECHO), Medicare Benefits Schedule (MBS), Model of Care (MoC), National Disability Insurance Scheme (NDIS), Newborn Blood Spot Screening (NBS), Pharmaceutical Benefits Scheme (PBS), Rare Awareness Education Support and Training (RArEST), Rare Voices Australia (RVA), United Kingdom (UK), United Nations (UN).

Appendix B: Summaries of Action Plan Pillars

Action Plan Pillar 1: Awareness and Education Summary

Action Plan Pillar 2: Care and Support Summary

Action Plan Pillar 3: Research and Data Summary

rare voices
AUSTRALIA®