

Engaging with the rare disease community

A summary of the stakeholder consultation process involved in developing the National Strategic Action Plan for Rare Diseases.

A companion document to the National Strategic Action Plan for Rare Diseases

February 2020

We acknowledge Traditional Owners of Country throughout Australia and recognise the continuing connection to lands, waters and communities. We pay our respect to Aboriginal and Torres Strait Islander people; and to Elders both past and present.

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Introduction

The collaborative development of the National Strategic Action Plan for Rare Diseases (the Action Plan) and its companion documents was led by Rare Voices Australia (RVA), with funding assistance from the Australian Government Department of Health.

This document is a companion document to the Action Plan. It details the stakeholder consultation process undertaken by RVA in the preparation of the Action Plan. It articulates the reasons for the process; provides a summary of the activities undertaken; and explains how the stakeholder consultation process informed the development of the Action Plan. It also acknowledges the stakeholders consulted and thanks them for their valuable time and input.

Background

RVA is experienced in leading consultation on rare disease issues with a wide range of stakeholders. In 2014, RVA embarked on a Rare Disease National Plan Engagement Roadshow, consisting of Roundtable discussions with people living with a rare disease, health care, research, government and industry stakeholders, with sessions in Queensland, Victoria, South Australia, New South Wales and Western Australia. The discussions focused on the need for, barriers to, and drivers of, coordinated action on rare diseases and for a National Plan.

The key findings from the Roadshow were presented at the 2015 National Rare Disease Summit and informed the Summit's focus and themes. The collaborative outcome of the 2015 Summit was a *Communiqué to progress A National Rare Disease Plan*, which was subsequently endorsed by more than 170 organisations or individuals in the rare disease community¹.

In June 2017, the themes of the *Communiqué* were further developed into the key advocacy and policy document, *Call for a National Rare Disease Framework: 6 Strategic Priorities*. This document was critical in creating traction in rare disease policy reform. At the 2018 National Rare Disease Summit, the Minister for Health, the Hon Greg Hunt MP, announced funding for RVA to collaboratively develop and deliver the National Strategic Action Plan for Rare Diseases². The *Call for a National Rare Disease Framework: 6 Strategic Priorities* policy document also served as the theme of the Summit. Speakers presented on the 6 Strategic Priorities: Diagnosis, Access to Treatments, Data Collection, Coordinated Care, Access to Services and Research. This announcement and the early collaboration at the Summit created the enabling environment for the stakeholder consultation process undertaken in developing the Action Plan.

Overview of consultations for the Action Plan

RVA led the collaborative development of the Action Plan with a Steering Committee consisting of cross-sector representation that provided guidance throughout the process.

Between late 2018 and mid-2019, RVA undertook a stakeholder consultation process that included the following activities:

- externally facilitated Roundtable consultation;
- face-to-face stakeholder consultations in Sydney, Melbourne, Perth and Brisbane;
- face-to-face consultation with pharmaceutical industry representatives;
- online survey;
- videoconference and email correspondence with RVA's Scientific & Medical Advisory Committee (SMAC);
- videoconference and email correspondence with the Action Plan Steering Committee;

¹ Rare Voices Australia 2019. *Rare Disease Summit Communiqué*. Accessed from www.rarevoices.org.au/page/57/communique on 12 September 2019.

² Australian Government Department of Health 2018. *Australia's First National Rare Diseases Framework*. Accessed from beta.health.gov.au/ministers/the-hon-greg-hunt-mp/media/australias-first-national-rare-diseases-framework on 27 June 2019.

- interviews with representatives of state and territory governments (mix of face-to-face and teleconference meetings);
- additional contact with content and population group experts throughout the Action Plan development; and
- review by the Australian Government Department of Health, including targeted conversations with representatives from the Pharmaceutical Benefits Advisory Committee, the Office of Health Technology Assessment, and the Australian Institute of Health and Welfare.

The consultation process was undertaken in an iterative manner, with each subsequent consultation building on the findings that emerged in those prior.

Rationale

One of the principles that underpins the Action Plan is a *person-centred approach*. It was therefore of the utmost importance that the Action Plan was developed in partnership with the people who it will ultimately affect, and thus, strongly reflect their experiences and ideas for change. Throughout this consultation process, RVA was struck by the strong sense of collaboration by, and generosity of, the sector. The Action Plan was seen as a real opportunity for change among stakeholders.

Findings

Hearing from such a wide range of stakeholders across the country resulted in a significant amount of information being shared with RVA. While there was diversity, and sometimes difference, there were many commonalities among the range of rare disease stakeholders.

Following the externally facilitated Roundtable on the Action Plan held in February 2019, RVA worked with members of the Action Plan Steering Committee to develop a draft 'Plan on a Page', which proposed a Vision, Critical Enablers, Policy Pillars, Priorities, and underlying Principles of the draft Action Plan.

Subsequent face-to-face consultations confirmed the core elements of this draft 'Plan on a Page', albeit with further refinement and greater nuance. These consultations also expanded on the 'Plan on a Page' to develop Actions and associated Implementation activities to support the Priorities.

Targeted and expert input was then sought from a broad range of stakeholders in order to build on the Actions and Implementation activities. This included an online survey; consultations with RVA's SMAC and the Action Plan Steering Committee; interviews with representatives of state and territory governments; additional contact with content and population group experts and; review by the Australian Government Department of Health, including subject matter experts.

Together, the stakeholder consultations formed the core of the Action Plan.

The Action Plan has been developed by the rare disease sector, for the rare disease sector.

Discussion: Consultations and Findings

Phase 1: Face-to-face stakeholder consultations

Externally facilitated Roundtable consultation on the National Strategic Action Plan for Rare Diseases with RVA staff, Department of Health staff and the Action Plan Steering Committee

The Roundtable consultation was held on 26 February 2019, and involved RVA, the Department of Health, and the Action Plan Steering Committee, which includes representation from all rare disease stakeholder perspectives and expertise. This established the initial directions for the development of the Action Plan.

The Roundtable was externally facilitated by Communication Link and was broadly based around the themes presented in RVA's *Call for a National Rare Disease Framework: 6 Strategic Priorities*:

- Diagnosis;
- Access to Treatment;
- Data Collection;
- Coordinated Care;
- Access to Services; and
- Coordinated Research.

Working in small groups, participants reviewed each of these themes to identify the key actions for each. Each action was then weighted so that actions across the priority areas could be compared with each other to identify the top actions overall. Finally, participants were asked to individually select their 'number one' priority.

Through the Roundtable, four key areas of action had majority support. These four areas are listed below:

1. The establishment of **centres of rare disease clinical and research excellence**.

- These centres of excellence will be a key driver of integrated care for people living with a rare disease, including those with a suspected but undiagnosed rare disease.
- This was the most frequently selected 'number one' action area, consistent with the highest ranked area during the group weighting exercise.

2. **Fair and equitable access to diagnosis**, particularly through better clinical diagnosis pathways, funding and integrations with the centres for excellence.

- A focus on diagnosis will ensure that people living with a suspected but undiagnosed rare disease are systematically identified.

3. **Increased access to approved medicines** through reform of the Pharmaceutical Benefits Advisory Committee (PBAC) process.

- A pathway with greater flexibility for rare disease therapies will ensure that people living with a rare disease are able to access medicines with demonstrated clinical benefit that are funded under the Pharmaceutical Benefits Scheme (PBS) for more common conditions.

4. National improvement to the **health care coding** of rare disease.

- Increased and improved data collection and use will ensure that rare disease can be automatically identified and measured in Australia.

Other key areas identified by participants included:

- prioritised rare disease **research** through the Medical Research Future Fund (MRFF) and the National Health and Medical Research Council (NHMRC) Strategic Plan, and;
- increased consultation with the rare disease community to identify service gaps and how to better provide **equitable service access**.

Communication Link's Roundtable Report is available on the RVA website.

Face-to-face stakeholder consultations in Sydney, Melbourne, Perth and Brisbane

Following the Roundtable consultation, a series of stakeholder consultations were conducted, some of which were open and some of which were targeted sessions, as follows:

- Sydney – 26 February 2019;
- Melbourne – 12 March 2019;
- Perth – 25 March 2019; and
- Brisbane – 29 April 2019.

Across all sessions, RVA consulted with approximately 100 members of the rare disease community, many of whom were in turn representing members of their specific organisation. Further detail on attendees is provided in Appendix 1.

The consultations were broadly based around the themes presented in RVA's *Call for a National Rare Disease Framework: 6 Strategic Priorities*. It was discussed that simpler language and categorisation of the priorities / themes would appeal to a broader range of stakeholders. To obtain the most valuable information from the expertise of attendees at each consultation, attendees were first asked to share their priorities for rare disease, before more targeted discussions took place. The face-to-face stakeholder consultations were critical in determining that the Action Plan should be *person-centred*; have a strong focus on *equity of access*, and; *prioritise sustainable systems and workforce*. It was clear that, in order to achieve the shared Vision, the following would be needed:

- Stakeholder involvement and engagement;
- Collaborative governance and leadership;
- State, national and international partnerships; and
- High quality, comprehensive collection, and effective use, of rare disease data.

From these conversations, the Action Plan began to take shape through the emergence of key policy themes and ideas for implementation. These themes form the Pillars of the Action Plan:

- Awareness and Education
- Care and Support
- Research and Data

Information relating to Implementation was collated and then organised thematically. It was then supplemented with information gathered through an online survey, as well as through an evidence review, systematic linkage with aligned documents (i.e. the Asia-Pacific Economic Cooperation (APEC) Action Plan on Rare Diseases) and scanning of related initiatives.

Phase 2: Online stakeholder consultation

Overview

Stakeholders who were unable to attend face-to-face consultations were offered the opportunity to respond to an online survey via the RVA mailing list, which reaches 1,800 subscribers (as at 30 October 2019), and via the RVA Facebook page, which has 5,952 followers (as at 30 October 2019). The survey was posted via the Survey Monkey platform and was open for two weeks.

As described previously, stakeholder consultations were undertaken in an iterative manner, with each consultation activity building on the inputs gained from those before it. The online survey therefore presented the Priorities and Actions³ that had emerged through face-to-face consultations to that point; asked respondents for their perspective on key early implementation steps in relation to the Actions, and; gave respondents an opportunity to put forward additional Priorities and Actions of importance to them and / or the organisation they were representing.

Two hundred people responded to the survey. Approximately one third participated as people living with a rare disease, another third as a carer for a person / people living with a rare disease, and the remaining third as employees / volunteers for a rare disease advocacy organisation, a clinician / practitioner or researcher / academic working in the rare disease space, or as an employee in the rare disease industry (i.e. private sector).

Contribution of Survey Feedback to Development of Action Plan

The following provides a summary of the main themes derived from the Survey.

Pillar 1: Awareness and Education

- Respondents agreed that national coordination and leadership is a critical element in connecting the rare disease community with decision-makers, and it was suggested that RVA could play a greater role in this in future.
- The critical role of rare disease organisations was emphasised by respondents, although it was acknowledged that the sector would benefit from greater consistency and coordination in its ongoing role in awareness and education.
- There was strong support for a coordinated and collaborative approach to developing a rare disease workforce strategy, central to which is health professional education and awareness-raising. Health professional education was also seen as a critical element in strengthening rare disease care and support.
- There was strong agreement that systems can and should play a greater role in raising awareness about rare disease within frontline health services, as well as the importance of education for practitioners to equip them to make rare disease diagnoses. Many practical suggestions were made, including system prompts and the development of formally endorsed education programs.
- The importance of a person-centred approach to care and support was emphasised. Respondents agreed it was important that rare disease organisation communications raise awareness of available support services.

Pillar 2: Care and Support

- Development of a rare disease-specific integrated care model was seen as critical, with some respondents pointing out pockets of success. A collaborative approach, involving all stakeholders, was suggested. That people living with rare disease and their families are at the centre, and are actively engaged with a strong voice, was raised by many as being important.
- Respondents agreed that responsive care and support is critical for rare diseases, given the often rapidly changing and/or progressive nature of many peoples' conditions. The role of policy and programs in supporting the delivery of integrated support and care was also acknowledged.

³ NB: Actions were called 'Policy Directions' in the survey. They were renamed Actions at a later date.

- Regarding diagnosis, respondents felt strongly that people living with an (undiagnosed) rare disease have the right to equitable access to a range of diagnostic tools and tests, regardless of one's ability of pay or the location in which one lives. National coordination and leadership were again seen as critical to overcoming these issues.
- The need for effective collection and use of rare disease data was raised as central to effective delivery of care and support.
- Respondents felt strongly about the role of best-practice diagnostic pathways, supported by workforce education and standardised approaches (clinical guidelines). The importance of support, including genetic counselling, for people on a diagnostic journey was also highlighted.
- With regard to access to rare disease health technologies, respondents raised the need for flexible reimbursement pathways, and for collaboration between parties such as rare disease organisations and clinicians, to ensure equity of access.
- Many respondents highlighted the important role played by rare disease organisations in the care and support ecosystem. There was a consistent call for increased funding / resourcing for these organisations, as well as the implementation of guidelines and standards to ensure their high quality.

Pillar 3: Research and Data

- Respondents raised the need for increased investment into all types of rare disease research.
- The importance of rare disease data was again highlighted, with respondents calling for national consistency, coordination and collaboration to obtain maximum benefit. Rare disease registries, including drug-related registries, were raised as critical sources of rare disease data with great potential for increasing knowledge.
- Respondents raised current inequities in terms of accessing clinical trials, including by Aboriginal and Torres Strait Islander people. They highlighted the enthusiasm of the rare disease community to be involved in clinical trials, and called for greater national coordination to increase the ability of Australians living with rare disease to access clinical trials.
- Gaps in research that were highlighted by respondents included the transition between services (i.e. paediatric to adult services), including research on ageing people living with a rare disease, and the translation of genomics / genetics research into clinical care settings.
- Respondents highlighted the importance of high-quality rare disease research, of collaboration and interdisciplinary research teams, and of sharing resources in rare disease so as to maximise efficiency and cost-effectiveness.

Phase 3: Targeted consultations

Face-to-face consultation with pharmaceutical industry representatives

A targeted consultation was held with representatives from the rare disease pharmaceutical industry on 8 April 2019. Many of these representatives are also members of RVA's Roundtable of Companies, a group of pharmaceutical companies with a common interest in rare diseases and orphan drug development. This consultation, like all others, was strongly *person-centred*, in line with the principles underpinning the Action Plan. The priority topics for industry included diagnosis, access to health technology, research and data collection.

The industry perspective provided unique insight into the challenges faced by the rare disease pharmaceutical industry in improving the health and wellbeing of people living with a rare disease, including in relation to Health Technology Assessment and reimbursement of emerging rare disease health technologies; data collection and reporting, including registries, and; the role of industry in rare disease research. Further detail on attendees is provided in Appendix 1.

Videoconference and email correspondence with RVA's SMAC

A videoconference was held with RVA's SMAC on 16 May 2019. SMAC members had been provided with draft documentation two weeks earlier and were able to provide feedback and input on this documentation both via email and during the videoconference. Further detail on SMAC membership is provided in Appendix 1.

SMAC members were instrumental in:

- undertaking a review of contemporary evidence related to rare disease policy, and the priority areas that had emerged through earlier elements of the consultation process, further detail on which can be found in the Companion Document titled, *The evidence base of the National Strategic Action Plan for Rare Diseases*.
- providing systematic linkage of the Action Plan Priorities and Actions with aligned documents, including:
 - Asia-Pacific Economic Cooperation (APEC) Action Plan on Rare Diseases;
 - National Strategic Framework for Chronic Conditions;
 - National Aboriginal and Torres Strait Islander Health Plan 2013–2023;
 - WHO Global Action Plan for the Prevention and Control of Noncommunicable Diseases; and
 - Other pre-eminent rare disease plans and strategies globally, including those in Europe, the United Kingdom, Canada and the United States.
 - Environmental scanning to find activities, initiatives and programs related to the Actions laid out in the Action Plan, further detail on which can be found in the Companion Document titled *Rare disease activities in Australia*.

Individual Committee members were also requested to provide advice relating to their areas of expertise, via email or phone. These conversations were related to Priorities, Actions or Implementation activities with high levels of technical detail, such as those related to data collection, genetic testing, or the research lifecycle.

Videoconference and email correspondence with the Action Plan Steering Committee

Further to the initial input provided by the Action Plan Steering Committee at the Roundtable Consultation, a videoconference was held on 30 May 2019 to provide members with an update of the Action Plan development process to date. Action Plan Steering Committee members had been provided with draft documentation one week earlier and were able to provide input and feedback on this documentation both via email and during the videoconference. At various points throughout the development of the Action Plan, individual Committee members were also requested to provide advice relating to their areas of expertise, via email or phone.

Input and feedback given by the Action Plan Steering Committee was then incorporated through a 'reconciliation process' against the Action Plan policy material, resulting in finessing of the material to incorporate key messages. Further detail on the Action Plan Committee membership is provided in Appendix 1.

Interviews with representatives of state and territory Governments (mix of face-to-face and teleconference meetings)

Much of the implementation of health, disability, social and other policy areas relevant to rare diseases occurs at the state and territory level. RVA undertook targeted consultations with representatives from state and territory governments as part of the Action Plan development process.

The Australian Government Department of Health provided RVA with a suggested contact within each jurisdiction, aside from Western Australia, for which RVA had existing rare disease policy contacts and established relationships. (Further detail is provided in Appendix 1.)

During interviews, representatives were asked to apply their policy and program expertise in reviewing the Action Plan policy material, as well as to provide information on rare disease policies, programs or initiatives in their jurisdiction.

An overarching theme to emerge from these interviews was that rare disease policy expertise is not embedded systematically throughout Australian governments. This presented a significant challenge to facilitating these interviews. However, the key themes to emerge are summarised below:

- the Action Plan presents an opportunity for real change in rare disease;
- the rare disease policy area requires national collaboration, as opposed to siloed jurisdictional implementation, due to the complexity and diversity of rare diseases paired with the small numbers in rare disease. Further to this:
 - it is important that the extensive overlap between rare disease and disability is given adequate consideration, in the context of current access issues experienced by people living with rare disease;
 - it is critical that rare disease data be captured in national data infrastructure, such as registries, including for undiagnosed rare disease. The significance of this is increasing with the rising prominence of genomics and precision / personalised medicine; and
 - the demand for genomics services is increasing, placing significant pressure on systems to meet demand and adequately support people on their genomic and / or diagnostic journey.
- it is important that the Action Plan aligns with relevant international, national and state/territory policies etc., in order to maximise alignment and minimise duplication. In particular, it is important that the Action Plan align with the National Health Genomics Policy Framework and Implementation Plan 2018–2021;
- it is important that the Action Plan and any subsequent implementation decisions are elevated through existing national and state/territory collaborative structures, such as through the Council of Australian Governments (COAG) Health Council (CHC), its advisory body the Australian Health Ministers' Advisory Council (AHMAC), its Clinical Principal Committee (CPC) and all other relevant governance structures;
- there are many opportunities to improve the care for people living with a rare disease. Care and support that is integrated and facilitated by interdisciplinary teams, is crucial in the rare disease context to overcome the complexity of such conditions. The current funding model, in which care is counted in Weighted Activity Units, and services are purchased accordingly is not conducive to true interdisciplinary collaboration;
- an Undiagnosed Disease Program in every jurisdiction is a critical part of rare disease infrastructure;
- it will be important to seek further and more targeted clinician input in developing implementation plans associated with the Action Plan, including those clinicians with expertise in relation to Priority Populations; and
- many jurisdictions shared opportunities for continuing collaboration.

Review by Federal Government Department of Health

The draft Action Plan was submitted to the Department of Health for initial review on 1 July 2019. After preliminary considerations by the Department, a revised (streamlined) version was distributed internally to relevant teams within the Department of Health for more detailed review. Collated feedback was then provided to RVA for consideration.

As a part of this stage, RVA sought input from, and held targeted conversations with, representatives from the Pharmaceutical Benefits Advisory Committee, the Office of Health Technology Assessment, and the Australian Institute of Health and Welfare (AIHW) in relation to specific implementation activities in which these areas were identified. A summary of these conversations is provided below:

Review by RVA Stakeholders

The revised draft National Strategic Action Plan for Rare Diseases was distributed to RVA stakeholders on 29 July 2019 as an opportunity to provide feedback. RVA collated all feedback received. These stakeholders included:

- all attendees of the externally facilitated Roundtable consultation and face-to-face consultations;
- RVA Partner organisations;
- state-based and national peak organisations representing people with genetic, undiagnosed and rare diseases;
- the Action Plan Steering Committee;
- RVA's SMAC;
- attendees at the Industry Consultation with representatives from the rare disease pharmaceutical industry, many of whom are also members of RVA's Roundtable of Companies;
- the representatives of state and territory governments with whom RVA had consulted; and
- other key stakeholders with expertise in certain areas covered within the Action Plan, including Research Australia, Consumers Health Forum of Australia, Medicines Australia and the Rare Disease International Working Group.

Conclusion

This document has demonstrated the invaluable contributions of each stakeholder consultation to developing not only the key elements of the Action Plan, which are outlined on the 'Plan on a Page', but also to the many Implementation activities laid out within the Action Plan.

Together, the stakeholder consultations truly formed the core of the Action Plan.

The Action Plan has been developed by the rare disease sector, for the rare disease sector.

Strong stakeholder engagement and consultation will continue to be of paramount importance as the Action Plan is implemented. The rare disease sector, including all identified implementation partners, have a unique opportunity to drive implementation of the Action Plan and to bring meaningful, positive and long-lasting change to the lives of Australians living with a rare disease, and their families and carers.

Appendix 1: Acknowledgements

Development of the Action Plan was led by RVA with funding from the Australian Government Department of Health.

Development of the Action Plan took place between late 2018 and 2019. Many individuals and organisations contributed time and expertise to the development of the Action Plan, including people living with a rare disease and their families or carers; rare disease organisations; rare disease health professionals; rare disease researchers; policy-makers, representatives from the Australian and state and territory government Departments of Health; and the rare disease pharmaceutical industry.

RVA sincerely thanks those who participated in the consultation and development phase, and acknowledges all who were able to attend consultations or provide input below.

Please note that the names of individual advocates or rare disease organisation leaders who attended consultations are not included below out of consideration for their privacy.

26 February 2019 – Sydney

Roundtable on National Strategic Action Plan for Rare Diseases, facilitated by Helen Leayr, Communication Link

Face-to-face meeting with one teleconferencing in

Communication Link

- Helen Leayr (Facilitator)

Rare Voices Australia

- Nicole Millis
- Sarah Cannata
- Anne Hunter
- Amy Mills

Action Plan Steering Committee

- Associate Professor Yvonne Zurynski – Associate Professor, Macquarie University
- Amanda Samanek – Executive Director, Genetic and Rare Disease Network
- Cameron Milliner – Asia Pacific Economic Cooperation (APEC) Life Sciences Innovation Forum (LSIF) Rare Disease Network Industry Co-Chair and former Head of Public Affairs and Patient Advocacy Asia Pacific, Takeda
- Monica Ferrie – Chief Executive Officer, Genetic Support Network of Victoria
- Heather Renton – Chief Executive Officer, Syndromes Without A Name (SWAN) Australia
- Clare Stuart – General Manager, Tuberos Sclerosis Australia (formerly)
- Greg Pratt – Aboriginal & Torres Strait Island Health Research Manager, QIMR Berghofer Medical Research Institute
- Associate Professor Carol Wicking – Independent Consultant
- Tiffany Boughtwood – Manager, Australian Genomics Health Alliance
- Kara Hunt – Grants Program Manager, Steve Waugh Foundation
- Eliza Mitchell – Manager – Policy and Public Affairs, Rare Cancers Australia

Department of Health (Commonwealth) representatives

Afternoon session

Face-to-face meeting

Individual Advocates and Rare Disease Organisation Leaders

- Parenteral Nutrition Down Under

- Australian Pompe Association
- ausEE
- Sanfilippo Children's Foundation
- Immune Deficiencies Foundation Australia
- Mito Foundation
- MPN Alliance Australia (Myeloproliferative Neoplasms)
- MdDS Australia (Mal de Debarquement Syndrome)
- Australian Addison's Disease Association Inc.
- Prader-Willi Research Foundation Australia

12 March 2019 – Melbourne

Face-to-face meeting

Rare Voices Australia

- Nicole Millis
- Sarah Cannata
- Anne Hunter
- Amy Mills
- Kathryn Milne

McKell Institute

- Angela Jackson (Facilitator)
- Edward Cavanough

Action Plan Steering Committee

- Simon McErlane – Medical Director Asia Pacific, Amicus Therapeutics
- Monica Ferrie – Chief Executive Officer, Genetic Support Network of Victoria
- Heather Renton – Chief Executive Officer, SWAN Australia
- Clare Stuart – General Manager, Tuberous Sclerosis Australia (formerly)
- Kara Hunt – Grants Program Manager, Steve Waugh Foundation

Individual Advocates and Rare Disease Organisation Leaders

- Batten Disease Support and Research Association
- Sanfilippo Children's Foundation
- PCD Australia (Primary Ciliary Dyskinesia)
- Immune Deficiencies Foundation Australia
- Porphyria Association Inc
- Angelman Syndrome Association Australia Inc
- Mito Foundation
- Prader-Willi Research Foundation of Australia
- Genetic Support Network Victoria
- Brotherhood of St Laurence (Local Area Coordinator for the National Disability Insurance Scheme)

Health Professional

- Queensland Lifespan Metabolic Medicine Service, Queensland Children's Hospital

25 March 2019 – Perth

Face-to-face meeting

Rare Voices Australia

- Nicole Millis
- Sarah Cannata
- Anne Hunter
- Kathryn Milne

Action Plan Steering Committee

- Amanda Samanek – Executive Director, Genetic and Rare Disease Network

Individual Advocates and Rare Disease Organisation Leaders

- Muscular Dystrophy WA
- Australian Pompe Association
- Angelman Syndrome Association Australia
- Australian Pituitary Foundation
- Genetic and Rare Disease Network
- Developmental Disability WA

Department of Health (WA)

- Office of Population Health Genomics
- Western Australian Register of Developmental Anomalies
- Genetic Services WA, North Metropolitan Health Service

Adaptive Process Services Pty Ltd

8 April 2019 – Sydney

Targeted Industry Consultation

Face-to-face meeting with two joining by teleconference

Rare Voices Australia

- Nicole Millis
- Sarah Cannata
- Kathryn Milne

Action Plan Steering Committee

- Simon McErlane – Medical Director Asia Pacific, Amicus Therapeutics

Industry Representative

- Alexion
- Amicus Therapeutics
- Biogen (teleconference)
- BioMarin
- Menarini
- Roche
- Sanofi

29 April 2019 – Brisbane

Face-to-face meeting

Rare Voices Australia

- Nicole Millis
- Sarah Cannata
- Anne Hunter
- Kathryn Milne

Action Plan Steering Committee

- Associate Professor Carol Wicking – Independent Consultant

Individual Advocates and Rare Disease Organisation Leaders

- ausEE
- Foundation for Angelman Syndrome Therapeutics Australia
- Immune Deficiencies Foundation Australia
- Metabolic Dietary Disorders Association
- Muscular Dystrophy Queensland
- Myasthenia Alliance Australia
- Myasthenia Gravis Association of Queensland Inc.
- Narcolepsy Australia
- Self Help Queensland
- Sleep Disorders Australia

Health Professionals

- Queensland Lifespan Metabolic Medicine Service, Queensland Children's Hospital
- Paediatric Neurology, Queensland Children's Hospital
- Emergency Department (Paediatrics), Queensland Children's Hospital

Queensland Genomics Health Alliance

May 2019

Online stakeholder consultation, in which 200 members of the rare disease community participated, including people living with a rare disease, people caring for someone living with a rare disease, people who work / volunteer for a rare disease advocacy organisation, clinicians / practitioners or researchers / academics working in the rare disease space, and people working in the rare disease industry (i.e. private sector).

16 May 2019 – RVA's SMAC

Videoconference

NB: RVA's SMAC was also approached for input and feedback via email at various points during the development of the Action Plan. Individual Committee members were also requested to provide advice relating to their areas of expertise, via email or phone.

- Associate Professor Carol Wicking (Chair) – Independent Consultant
- Associate Professor Gareth Baynam – Associate Professor, Western Australian Health Department
- Professor Alan Bittles – Adjunct Professor and Research Leader, Murdoch University and Adjunct Professor, Edith Cowan University
- Dr Lisa Ewans – Clinical Geneticist, Royal Prince Alfred Hospital and Clinical Associate Lecturer, The University of Sydney
- Professor Adam Jaffe – Head of the Discipline of Paediatrics, University of NSW, Associate Director of Research, Sydney Children's Hospitals Network
- Dr Paul Lacaze – Head of Public Health Genomics at Monash University

- Dr Kristen Nowak – Director of the Office of Population Health Genomics, Western Australian Health Department
- Dr Lemuel Pelentsov – Program Director, University of South Australia, School of Nursing and Midwifery
- Professor Jeff Szer AM – Director, Royal Melbourne Hospital
- Associate Professor Yvonne Zurynski – Associate Professor, Macquarie University
- Dr Kaustuv Bhattacharya – Metabolic Specialist, Queensland Lifespan Metabolic Medicine Service, Queensland Children’s Hospital

30 May 2019 – National Strategic Action Plan for Rare Diseases Steering Committee

Videoconference

NB: The National Strategic Action Plan for Rare Diseases Steering Committee was also approached for input and feedback via email at various points during the development of the Action Plan. Individual Action Plan Steering Committee members were also requested to provide advice relating to their areas of expertise, via email or phone.

Steering Committee Members:

- Associate Professor Gareth Baynam – Associate Professor, Western Australian Health Department
- Cameron Milliner –APEC LSIF Rare Disease Network Industry Co-Chair and former Head of Public Affairs and Patient Advocacy Asia Pacific, Takeda
- Tiffany Boughtwood – Manager, Australian Genomics Health Alliance
- Monica Ferrie – Chief Executive Officer, Genetic Support Network of Victoria
- Kara Hunt – Grants Program Manager, Steve Waugh Foundation
- Professor Adam Jaffe – Head of the Discipline of Paediatrics, University of NSW, Associate Director of Research, Sydney Children’s Hospitals Network
- Simon McErlane – Medical Director Asia Pacific, Amicus Therapeutics
- Bryan McDade – Patient Care Co-ordinator, Rare Cancers Australia
- Jan Mumford – Executive Director, Genetic Alliance Australia
- Greg Pratt – Aboriginal & Torres Strait Island Health Research Manager, QIMR Berghofer Medical Research Institute
- Heather Renton – Chief Executive Officer, SWAN Australia
- Amanda Samanek – Executive Director, Genetic and Rare Disease Network
- Clare Stuart – General Manager, Tuberous Sclerosis Australia (formerly)
- Associate Professor Carol Wicking – Independent Consultant
- Associate Professor Yvonne Zurynski – Associate Professor, Macquarie University

State and Territory Departments of Health

Mix of telephone and face-to-face interviews

Australian Capital Territory – Health Directorate – representatives

New South Wales – Agency for Clinical Innovation – representatives

Northern Territory – Department of Health – representatives

Queensland – Queensland Health

South Australia – Department of Health and Wellbeing – representatives

Tasmania – Department of Health – representatives

Victoria – Department of Health and Human Services – representatives

Western Australia – Department of Health – representatives

Australian Government

Mix of email and face-to-face input and feedback

Pharmaceutical Benefits Advisory Committee – representatives

Office of Health Technology Assessment – representatives

Australian Institute of Health and Welfare – representatives

A number of areas within the Department of Health were provided a draft of the Action Plan and given the opportunity to provide feedback.

Review by RVA Stakeholders

RVA also distributed the draft Action Plan to key stakeholders as an opportunity to provide feedback. These stakeholders included:

- all attendees of the externally facilitated Roundtable consultation and face-to-face consultations;
- RVA Partner organisations;
- state-based and national peak organisations representing people with genetic, undiagnosed and rare diseases;
- the Action Plan Steering Committee;
- RVA's SMAC;
- RVA's Roundtable of Companies (pharmaceutical companies with a common interest in rare diseases and orphan drug development);
- the representatives of state and territory governments with whom RVA had consulted; and
- other key stakeholders with expertise in certain areas covered within the Action Plan, including Research Australia, Consumers Health Forum of Australia, Medicines Australia and the Rare Disease International Working Group.

