

The National Strategic Action Plan for Rare Diseases: A Collaborative Multi-stakeholder Approach to Effective Rare Disease Policy Reform

INTRODUCTION

Rare Voices Australia (RVA) led the collaborative development of the National Strategic Action Plan for Rare Diseases¹ (the Action Plan), the first coordinated effort to address rare diseases (RD) in Australia. The Action Plan was launched by the Minister for Health in February 2020 and addresses the many commonalities of RD across three interrelated Pillars: Awareness and Education; Care and Support; and Research and Data. It outlines a comprehensive, collaborative and evidence-based approach built on three principles: person-centred; equity of access; and sustainable systems and workforce.

AIM

A review of strengths and learnings from the collaborative multi-stakeholder development of the Action Plan and how this approach has impacted RD policy reform in Australia.

METHODS

The Action Plan is a case study of national policy development guided by a Steering Committee consisting of cross-sectoral (multi-stakeholder) representation. Between late 2018 and mid-2019, multiple methods of qualitative data collection/analysis was used through a national stakeholder consultation. Discussion themes were based on RVA's six strategic priorities: diagnosis, access to treatments, data collection, coordinated care, access to services and research. The consultation process was conducted in an iterative manner, with each subsequent consultation building on previous findings.

PHASE 1a – Face-to-face Steering Committee Roundtable

Roundtable consultation with multi-stakeholder Action Plan Steering Committee, RVA and Federal Department of Health staff. In small groups, participants identified the key actions for each priority, which were then weighted across each priority area to identify the top actions overall. Finally, participants were asked to individually select their 'number one' priority.

PHASE 1b – Face-to-face multi-stakeholder consultations

Four consultations (Sydney, Melbourne, Perth, Brisbane) involving 100 participants across the RD sector. Of the attendees, 80% were patient group/organisation leaders representing their community resulting in a strong person-centred focus. Attendees were asked to share their priorities for RD ahead of targeted discussions.

PHASE 2 – Online survey

Other stakeholders had the opportunity to respond to an online survey (Survey Monkey platform) open for two weeks via the RVA mailing list and social media channels. Two hundred people responded. Approximately one third were a person living with a RD; one third were a carer of a person with a RD; and the remaining third were a RD organisation leader, clinician, researcher, or industry stakeholder. The survey presented priorities and actions that had emerged so far through consultations and asked respondents for their perspective on early Action Plan implementation. Respondents also offered additional priorities and actions of importance to them or the organisation they represented.

PHASE 3 – Targeted consultations

- Face-to-face consultation with pharmaceutical industry representatives. Discussion focused on diagnosis, access to treatments, data collection and clinical trials.
- Videoconference and email correspondence with RVA's Scientific & Medical Advisory Committee (SMAC) and the Action Plan Steering Committee. Review of draft Action Plan documentation. As required, individual Steering Committee members were asked to provide advice relating to their areas of expertise via email/phone.
- Interviews with representatives of state and territory governments (face-to-face and teleconference). Representatives were asked to apply their policy/program expertise to review the Action Plan material, and to provide information on RD policies, programs or initiatives in their jurisdiction.
- Additional contact with content and population group experts.
- Discussions with representatives from the Pharmaceutical Benefits Advisory Committee (PBAC), the Office of Health Technology Assessment, and the Australian Institute of Health and Welfare.

PHASE 4 – Review of Action Plan draft (RVA stakeholders)

Stakeholders included all consultation attendees; RVA Partner organisations: state and national peak organisations representing people with genetic, undiagnosed and RD; the Action Plan Steering Committee; RVA's SMAC; attendees from the industry consultation; representatives of state and territory governments with whom RVA had consulted; and other key stakeholders (Research Australia, Consumers Health Forum of Australia and Medicines Australia).

PHASE 5 – Review of the Action Plan draft by Australian Government Department of Health

RESULTS

PHASE 1a – Face-to-face Steering Committee Roundtable

Four key areas had majority support:

1. Establishment of centres of RD clinical/research excellence.
2. Equitable access to diagnosis and better clinical pathways.
3. Improved access to medicines through reform of approval processes (e.g. PBAC).
4. National health care coding of RD/data collection for better RD identification/measurement.

Other areas identified: need for prioritised RD research and increased consultation with the RD community to identify/address gaps and inequities.

PHASE 1b – Face-to-face multi-stakeholder consultations

The face-to-face stakeholder consultations helped to shape the Action Plan's vision, principles and critical enablers. From these conversations, the Action Plan began to take shape through the emergence of key policy themes and ideas for implementation.

PHASE 2 – Online survey

Summary of the main themes from the online survey.

Pillar 1: Awareness and Education

- Need for national coordination and leadership; greater role for RVA as the peak body.
- The critical role of RD organisations, although they need greater consistency and coordination.
- Strong support for coordinated collaborative development of a RD workforce strategy.
- Need for increased awareness of RD within frontline health services to assist timely diagnosis including system prompts and formal education programs.

Pillar 2: Care and Support

- Collaborative person-centred development of a RD-specific integrated care model is critical.
- Needs to be responsive and timely due to the often changing/progressive nature of many RDs and supported by policy and program delivery.
- Equitable access to a range of diagnostic tools and tests. National coordination and leadership were seen as critical.
- Effective collection and use of RD data is central to effective service delivery.
- The need for best-practice diagnostic pathways, supported by workforce education and clinical guidelines. The importance of support for people on a diagnostic journey was also highlighted.
- The need for flexible reimbursement pathways and better stakeholder collaboration to ensure equity of access.
- Need for increased funding/resourcing for RD organisations in recognition of their central role.

Pillar 3: Research and Data

- Need for increased investment into all types of RD research.
- Need for national consistent, coordinated and collaborative data collection (e.g. RD registries).
- Current inequities in accessing clinical trials. Need for greater national coordination.
- Highlighted research gaps including transition between services (e.g. paediatric to adult services), including research on ageing people living with a RD, and the translation of genomics research into clinical care settings.
- Need for collaboration and interdisciplinary research teams, and sharing resources to maximise efficiency and cost-effectiveness.

PHASE 3 – Targeted consultations

Consultation with governments

An overarching theme to emerge from these interviews was that RD policy expertise is not embedded systematically throughout Australian governments. A telling example of this is that it was often unclear who was responsible for RD policy and who RVA should interview within each jurisdiction/government department.

Key themes from targeted consultations:

- The Action Plan presents an opportunity for real change in RD.
- National collaboration is required to overcome jurisdictional silos.
- There is extensive overlap between RD and disability.
- Need for national data infrastructure such as registries, including for undiagnosed RD.
- Increasing demand for genomics and precision/personalised medicine, which places significant pressure on current systems.
- The Action Plan must align with relevant international, national and state/territory policies, in order to maximise alignment and minimise duplication.
- Important that the Action Plan and implementation are elevated through existing national and state/territory collaborative government structures.
- Integrated care and support by interdisciplinary teams is crucial for the complexity of RD.
- An Undiagnosed Disease Program in every jurisdiction is critical.

DISCUSSION

National stakeholder consultation resulted in a significant amount of information from various perspectives. While there was diversity, there were many commonalities among RD stakeholders. The commonalities identified through the stakeholder consultations guided the Action Plan.³



A review of this policy development process highlights the importance of collaboration and multi-stakeholder engagement. Collaboration and multi-stakeholder engagement continues to shape initial implementation and policy reform in response to the Action Plan.

CASE STUDY: COLLABORATIVE MULTI-STAKEHOLDER POLICY REFORM OUTCOMES

The Action Plan helped to drive the establishment of the Parliamentary Inquiry into approval processes for new drugs and novel medical technologies in Australia, with a particular focus on access to the treatment of RD and conditions where there is high and unmet clinical need. The Inquiry was announced in August 2020 and due to COVID-19, public hearings are expected to be scheduled for 2021.

"We're looking forward to hearing from the public, health professionals and organisations, pharmaceutical companies and other interested parties on how Australia could position itself best on providing access to new drugs and novel medical technologies for all Australians now and into the future." – Chair, Standing Committee on Health, Aged Care and Sport, Mr Trent Zimmermann MP⁴

CONCLUSION

The Action Plan is an effective case study of collaborative and multi-stakeholder policy development. Stakeholder consultation was central to the Action Plan's development and formed the core of the Action Plan. The Action Plan has been developed 'by the rare disease sector, for the rare disease sector'. Strong multi-stakeholder engagement and consultation will continue to be of paramount importance as the Action Plan is implemented. The RD sector has "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."⁵



Minister for Health, Hon Greg Hunt MP, launching the National Strategic Action Plan for Rare Diseases.

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