RArEST
Rare Disease Awareness, Education, Support, and Training

Taking Action Together Toolkit

Improving Health and Wellbeing Outcomes for Australians Living with a Rare Disease

Acknowledgement of Country

We acknowledge and pay respects to all Aboriginal and Torres Strait Islander peoples across Australia, past and present. We acknowledge this document was designed and produced on Aboriginal and Torres Strait Islander land. We acknowledge the Traditional Owners of Country and recognise their continuing connection to lands, waters, and communities.

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University of Western Australia

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Endorsed by:

Contents

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Rare disease on a page

What is a rare disease?

A rare disease is a chronic and complex condition that affects **less than five in 10,000** people.[1,2]

How many rare diseases are there?

Over **7,000 known rare diseases** exist with more yet to be named. Unknown or unclassified diseases are referred to as *undiagnosed diseases*.[1,2]

What are the types of rare diseases?

80% are of genetic origin although rare diseases also include rare cancers, infections, post-infection conditions and autoimmune diseases.[3,4]

How many Australians live with a rare disease?

An estimated two million Australians currently live with a reare disease which is **approximately 8% of Australians**.[5]

Figure 1. Key facts about rare diseases[1-5]

The challenges of living with a rare disease

People living with a rare disease face many challenges. Rare diseases often first develop during childhood and most are long-term (chronic) and complex.[5-12] People often experience:

- Difficulty accessing health care
- Shortened lifespans
- High care needs
- Disability
- Limited care coordination
- Difficulty getting referrals to specialist health professionals
- Financial hardships due to out-of-pocket health care costs
- Ongoing stress, uncertainty, loneliness, and grief.

To read more about rare diseases in Australia visit:

- **Rare Voices Australia (RVA) website.** Information and resources from RVA, the national peak body for Australians living with a rare disease: https://rarevoices.org.au.
- **Rare Awareness Rare Education (RARE) Portal (RVA).** Contains current, reliable, and straightforward information and resources for all rare disease stakeholders customised for the Australian context: https://rareportal.org.au.
- **Online Education Portal (RVA).** Rare disease resources and courses: https://education.rarevoices.org.au.
- **What we're doing about rare diseases (Department of Health and Aged Care).** Information about Federal Government rare disease initiatives: https://www.health.gov.au/topics/chronic-conditions/what-were-doing-about-chronic-conditions/what-were-doing-about-rare-diseases.

Section 1

About this Toolkit

The Australian Government's National Strategic Action Plan for Rare Diseases (the Action Plan), was launched in 2020. The Action Plan is the first nationally co-ordinated effort to address rare disease in Australia.[13]

RVA led the collaborative development of the Action Plan, which outlines the priorities, actions, and implementation steps required to achieve the best possible health and wellbeing outcomes for Australians living with a rare disease.

This Toolkit aligns with the Action Plan, which is centred around three interrelated pillars, as shown in Figure 2, below. More information about the Action Plan can be found on RVA's website.

Figure 2. The three Pillars of the National Strategic Action Plan for Rare Diseases

The Action Plan highlights the importance and value of involving and engaging many different people and organisations (stakeholders) in actions and activities to improve health outcomes and experiences of Australians living with rare disease. **Rare disease stakeholders** include:

- People living with a rare disease, including those with an undiagnosed rare disease, families and carers
- Consumer-led organisations, peak bodies, and charities
- Health professionals
- Academics, researchers, and research funders
- Governments and policy makers
- Biotechnology and pharmaceutical companies.

These different key stakeholders make up the **rare disease sector** (Figure 3). More details about these groups can be found in the Toolkit companion document *Who's who: the people and organisations involved in the Australian rare disease sector*.

About this Toolkit (continued)

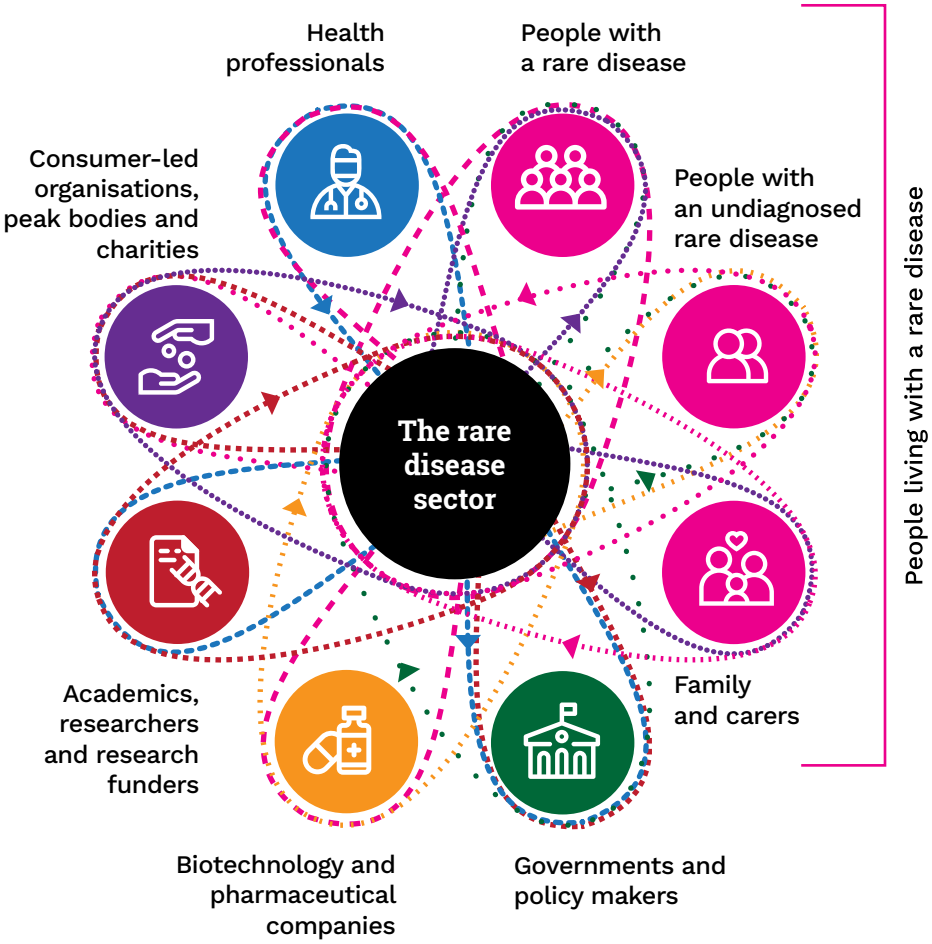


Figure 3. Key members of the rare disease sector

What the Toolkit is for

This Toolkit demonstrates how the Australian rare disease sector has worked effectively together to **advocate** (speak up) for **systemic change** benefitting the rare disease community. **Systemic changes** are those that result in changes to the systems involved in providing care and support to people living with a rare disease.

The Toolkit describes key approaches that people and organisations have taken that resulted in **effective systemic advocacy and positive change** for Australians living with a rare disease.

These changes have improved the health and wellbeing of Australians living with rare disease by enhancing:

- awareness and education,
- care and support, and
- research and data.

Effective approaches are ones that gain and maintain the trust of all stakeholders, and lead to a common desired outcome. Trust among all members of the rare disease sector is important, and gaining and maintaining the trust of the rare disease community is crucial to effective systemic advocacy and positive change.

About this Toolkit (continued)

The Toolkit is designed to help people:

- Learn about approaches that can help to bring about systemic change
- Plan their own approaches and next steps

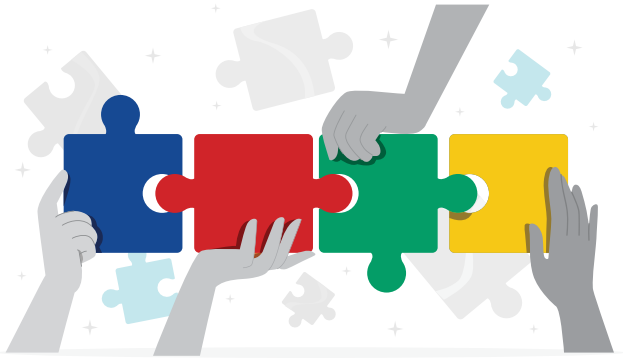
Although the rare disease sector extends beyond health to the disability, social and welfare sectors, education, employment, housing and other areas, this Toolkit primarily focuses on the **health system**, including health policy at state and federal levels, health service provision, medical and health research, clinical trials, access to therapies and clinical care.

Who the Toolkit is for

To help address the challenges faced by people living with a rare disease, the Toolkit provides inspiration, resources, practical tools and links to training and education to help the **entire rare disease sector** advocate more effectively to achieve the best possible health and wellbeing outcomes for people living with a rare disease.

How to use the Toolkit

Section 2 includes case studies provided by rare disease organisations and rare disease advocates which are designed to showcase how different stakeholders have worked together to bring about systemic change.



These case studies highlight **four key approaches** commonly used by stakeholders to achieve systemic change in rare disease (Table 1). The intention is that you can read these case studies to gain inspiration and insight into approaches that have been effective.

Then, in Section 3, each approach is described in more detail. Accompanying tools are provided that could be helpful for individuals and organisations considering how to apply these approaches to their own systemic advocacy.

About this Toolkit (continued)

Table 1. The key approaches for effective systemic advocacy described in this Toolkit

Networking and collaboration	How to find your allies and experts
Communication and storytelling	How to create clear messaging and share personal stories safely
Co-design	How to apply co-design to ensure the needs and priorities of all rare disease stakeholders are included
Patience, persistence, and flexibility	How to keep going despite the uncertainty and challenges – rare disease challenges are complex and often require complex solutions

The icons below are used throughout the Toolkit:

- Rare disease-specific tools
- Links to more information and practical tools
- Quotes from the rare disease community
- Key messages
- Print

How the Toolkit was developed

This Toolkit was developed as part of the Australian Government-funded Rare Disease Awareness, Education, Support and Training (RArEST) Project. The RArEST Project is a partnership between RVA, the University of New South Wales (UNSW Sydney), Macquarie University, and the University of Western Australia.

The Toolkit was co-developed with the rare disease sector, including people living with a rare disease, leaders of rare disease organisations, health professionals, and RVA as the national peak body for Australians living with a rare disease. More information about the development process and the frameworks that guided the Toolkit development can be found in the Toolkit companion document *How the Toolkit was developed*.

Section 2

Case Studies

The Australian rare disease sector has a strong history of working together to drive systemic change. The case studies below give examples of effective advocacy, including the challenges encountered.

They highlight four common approaches - **networking and collaboration**, **communication and storytelling**, **co-design**, and **patience, persistence, and flexibility**. These approaches are explored in more detail in Section 3.

Click on the boxes below to explore the case studies in more detail.

Case Study 1: Rare Voices Australia

On behalf of the Australian Government, Rare Voices Australia led the collaborative development of the National Strategic Action Plan for Rare Diseases.

"... partnership with Australians living with a rare disease was paramount and helped identify the goals and actions of the Action Plan."

Case Study 2: Mito Foundation

The Mito Foundation in partnership with their community, successfully advocated for the legislation of mitochondrial donation in Australia, resulting in the Australian Parliament passing the Mitochondrial Donation Law Reform (Maeve's Law) Act 2022.

"...the community were both the heart and engine room of the campaign..."

Case Study 3: Sydney Children's Hospitals Network and the University of NSW

A clinical team at Sydney Children's Hospitals Network established and integrated clinical trials into routine care for people living with spinal muscular atrophy.

"Clear evidence of better health outcomes and stories reflecting lived experience was a powerful combination for setting advocacy goals."

Case Study 4: SATB2 Connect

SATB2 Connect partnered with researchers at four universities to co-design research papers on *SATB2* Associated Syndrome.

"Our families are the experts and the co-designers of the research."

Case Study 1

Leading the collaborative development of the Australian Government's National Strategic Action Plan for Rare Diseases

The National Strategic Action Plan for Rare Diseases (the Action Plan) is the first nationally coordinated effort to address rare diseases in Australia.[13] RVA was commissioned by the Australian Government to lead the collaborative development of the Action Plan. Informed by an extensive multistakeholder consultation process, the Action Plan represents the views of the rare disease sector. It outlines a comprehensive, collaborative, and evidence-based approach.

Our advocacy goal

As the national peak body for Australians living with a rare disease, our goal was to develop an Action Plan in partnership with the rare disease sector, for the rare disease sector. Launched in 2020, the Action Plan is a nationally coordinated, comprehensive policy framework that responds to the challenges of living with a rare disease and acknowledges rare diseases as a national policy priority.

The Action Plan's vision is to drive the best possible health and wellbeing outcomes for Australians living with a rare disease through collaboration and co-design within and by the Australian rare disease sector. It aligns Australian rare disease policy with international best-practice policy frameworks and provides a shared policy language for the rare disease sector.

Ensuring key voices in the rare disease sector were heard and considered

A primary principle of the Action Plan is a person-centred approach. Working in **partnership** with Australians living with a rare disease was key and helped identify goals and priorities within the Action Plan. We were struck by the entire sector's willingness to **collaborate**.

Approach 1: Networking and collaboration

Working in partnership and consulting with different stakeholders in the rare disease sector meant the Action Plan reflected a wide range of goals and priorities.

Case Study 1 (continued)

We listened to the **personal stories** of people living with a rare disease and other key stakeholders and asked for people's thoughts throughout the process. Although feedback from members of the rare disease sector sometimes differed, there were many commonalities. These commonalities were prioritised, and when differences of opinions persisted, we focused on what was valuable to most people living with a rare disease.

Approach 2: Communication and storytelling

By listening to the stories of people living with a rare disease, RVA were able to better understand the needs and priorities of their community.

We used an iterative approach, with each consultation building on the outcomes of prior consultations. Activities that ensured effective **co-design** of the Action Plan included externally facilitated roundtable consultations, face-to-face stakeholder consultations in multiple cities across Australia, and interviews with representatives from state and territory governments.

Partnerships, co-design, and collaboration were key to developing an Action Plan that generated multipartisan support

The Action Plan was **jointly developed** by the Australian rare disease sector and the Australian Government. The Action Plan has received multipartisan support, including support from the Australian Government and the opposition political party.

In response to the Action Plan, the Australian Government funded several projects, including the Rare Disease Awareness, Education, Support and Training (RArEST) Project. The Action Plan has also informed other Australian Government policy reviews and consultations, for example, the National Medicines Policy review, and the House of Representatives Standing Committee on Health, Aged Care and Sport's Parliamentary Inquiry into approval processes for new drugs and novel medical technologies in Australia.

Approach 3: Co-design

The Action Plan was developed 'by the rare disease sector, for the rare disease sector.'

Case Study 1 (continued)

Several organisations have used the Action Plan to guide their activities, including Western Australia's Health Department, Sydney Local Health District, the Garvan Institute of Medical Research, the Murdoch Children's Research Institute and Telethon Kids Institute.

The Action Plan is an example of **multi-stakeholder collaboration** and **co-design**, a shared language among the sector, and laying the foundations for better health and wellbeing outcomes for people living with a rare disease.

Key messages

This case study highlights the value of:

- **Networking and collaboration** to gain input and support from key stakeholders in the rare disease sector
- **Communication and storytelling** to ensure the voices and needs of people living with a rare disease were embedded into the Action Plan
- **Co-design** to ensure the Action Plan captured the priorities of the rare disease sector

"Although feedback from stakeholders sometimes differed, there were many commonalities. These commonalities were prioritised, and when differences of opinions persisted, we focused on what was most valuable to people living with a rare disease."

Case Study 2

Leading a national advocacy campaign to successfully change the law and improve access to a new health technology

Mitochondrial disease (mito) is a chronic and life-limiting condition caused by changes in the way the mitochondria (the 'powerhouses' of our cells) work.[14] These changes rob the body's cells of energy and can cause symptoms in any organ at any age. As a result, mito can have a major impact on people's daily lives and lead to early death.

Mitochondrial donation is an assisted reproductive technology that helps women with some forms of mito to avoid passing the condition on to their biological children.[15] The technique uses in vitro fertilisation with the aim of ensuring only healthy mitochondria are passed on. Before the new law permitting mitochondrial donation was introduced, the procedure was illegal in Australia. Consequently, women affected by the most common types of mito were unable to have biological children who did not have mito. The limited options available to them to have children without mito were to conceive using donor eggs, adopt, or foster.

Our advocacy goal

Mito Foundation is a not-for-profit organisation that supports people affected by mitochondrial disease (mito), funds essential research into the prevention, diagnosis, treatment and cure of mitochondrial disorders, and increases awareness and education about mito.[16]

We wanted to change Australian law to make mitochondrial donation legal. Mitochondrial donation was being explored in research and had been considered in an Australian legislative review in 2011. When mitochondrial donation was legalised in the United Kingdom, the Mito Foundation realised we could play a significant role in making a similar change in Australia.

Including this specific goal in our 2016 Strategic Plan was the start of real momentum. We stated that we wanted to "drive legislative change so that Australian patients can have access to mitochondrial donation within five years". We achieved this goal with the Mitochondrial Donation Law Reform (Maeve's Law) Bill 2021 being passed in the Australian Senate on 20 March 2022. This was a conscious vote in both houses of Parliament, which meant we needed to communicate with many different Federal politicians.

Case Study 2 (continued)

Clear and consistent communication: ensuring the lived experience and scientific expertise of the mito community was heard

Communication was a key part of our campaign. The mito community (those with lived experience of mitochondrial disease) were front and centre. We supported people to meet with their elected representatives and **share their stories**. Their advocacy was the fuel of our campaign.

We also engaged a professional Government Relations Advisor who worked closely with us and helped with building our skills throughout the campaign.

Our medical and scientific advisors and partners ensured we represented the science accurately and advocated within their own settings. One example was the Open Letter published in *The Canberra Times* in the week of the Senate vote, which was signed by 60 leading Australian scientists.

Approach 2: Communication and storytelling

Guiding the mito community in how to effectively share their stories helped Parliamentarians understand the need for a change in the Australian law.

Finding and engaging effectively with stakeholders and partners

Key strategies that worked well included using **existing networks of health professionals and researchers**, working directly with the **mito community**, and supporting them in their advocacy activities. We maintained a Target Stakeholder Activity Manager, which was used to track all interactions with stakeholders and stakeholder meeting documents, including briefs for Parliamentarians.

During this process, we **kept the mito community updated** through webinars and a dedicated website that provided updates on the campaign and housed resources they could use in their advocacy. We had speakers from the United Kingdom visit Australia and join online discussions to maintain interest in the topic and keep us, the mito community, and our stakeholders informed.

We learnt a lot from reform being implemented in the UK and we **shared what we learnt** with the mito community in Australia.

Approach 1: Networking and collaboration

By tracking all interactions with stakeholders and keeping the community updated, a wide, informed, and supportive network was built to help get Mito Foundation's message out across Australia.

Case Study 2 (continued)

Patient and persistent engagement to influence decision makers

Other key approaches were **patient and persistent relationship-building and management** through meetings with key political supporters, activating the mito community to **speak to their elected representatives**, engaging with a public relations agency that had a focus on political advocacy to guide our use of the media in the campaign, showing gratitude to all stakeholders at every stage and **knowing when to go hard and when to be patient** with the process. These strategies were informed by our Government Relations Advisor.

Less effective strategies were using online petitions and letter-writing tools.

Approach 4:
Patience, persistence and flexibility

Building and maintaining strong relationships with a range of stakeholders was a key approach. This was done in a patient but persistent way.

Some of the challenges we faced

- **Mitochondrial donation will not benefit all community members:** A significant proportion of the mito community have types of mito that will not benefit from mitochondrial donation. We made sure that our support services and research funding continued so that mitochondrial donation was not our sole focus. We also tried to use language that was supportive of choice, as we knew not all families would choose this option.
- **Being just public enough:** We wanted to build political support, but also avoid sensationalising the issue and unearthing objectors. We used the media selectively and took advice from our public relations partner to achieve this balance.
- **Working to a timeline we didn't control:** The Government set the process that would lead to the change, including two Senate inquiries and a consultation by the National Health and Medical Research Council (NHMRC). The COVID-19 pandemic led to progress being stalled. We had to revise our approach at each stage, identify ways we could contribute and keep the momentum going. For example, in November 2020 we asked the community to write to the Prime Minister and Health Minister calling for an announcement to ensure that another Christmas did not pass without progress. Work re-started in February 2021.

Case Study 2 (continued)

Our key 'take homes' for how we achieved our advocacy goal

- ✓ **Be bold.** It was overwhelming at first to take on the task of changing the law. But without taking on the challenge, we would not have succeeded.
- ✓ **Build and activate a community.** The community were both the heart and engine room of the campaign. We supported them to reach out to local representatives to build support for change and to identify champions as well as understand those who didn't support the law change.
- ✓ **Build and buy capabilities.** We knew that we didn't have the capabilities to do, and outsourced by engaging a professional Government Relations Advisor and a public affairs agency.
- ✓ **Collaborate.** We did not do this alone. We funded a citizens' jury project to generate evidence to support the change.[17] A citizens' jury is a group of lay citizens who hear evidence about and discuss a challenging issue. Each participant then contributes to a 'verdict'. In our citizens' jury, most jurors approved of mitochondrial donation becoming available in Australia. We also enlisted researchers and their organisations to pledge public support and be involved in various stages of the campaign.
- ✓ **Show gratitude.** We focused on this throughout the campaign with all stakeholders. We even expressed gratitude to those Parliamentarians who voted against the law for their robust debate of the issues!

Key messages

This case study highlights the value of:

- **Networking and collaboration** to gain support and expertise from across the rare disease sector
- **Communication and storytelling** to ensure the voices and needs of the mito community were heard by policymakers
- Being **patient and persistent** when building and maintaining relationships

"Many stakeholders and collaborators were essential to this campaign, primarily, the mito community... [who] were front and centre in the campaign."

Case Study 3

Developing and translating the first treatments for spinal muscular atrophy into clinical practice

Spinal muscular atrophy (SMA) is a genetic condition that causes weakness and wasting of the skeletal muscles, which are the muscles used for movement.[18-20] Signs of the most common form of SMA, type 1, usually appear within the first months of life, and many children do not survive past infancy. With all SMA types, muscle weakness generally worsens with age, and affects people's ability to sit, stand, and walk.

Our advocacy goal

Our goals were to:

- Conduct SMA clinical trials at the Sydney Children's Hospitals Network (SCHN) Randwick Neuromuscular Clinic and integrate trials into clinical care
- Prepare our health service for rapid incorporation of new treatments into routine clinical care, including quick and accurate diagnosis, ideally as part of newborn screening

Our first step to achieving these goals was being chosen to be part of an international clinical trial. To do this we had to show it was feasible to run a trial at SCHN. We had to show we had a standard approach of caring for our patients that was used across SCHN, and that SCHN had the right people and resources to run the trial.

We worked as a **Centre of Expertise** to provide multi-disciplinary care, that is, care that is provided by different types of health care professionals. A Centre of Expertise provides specialist care for people with specific conditions or groups of conditions (for example, neuromuscular conditions or rare diseases) and supports research, education and community engagement.

Building a community

One of our key approaches was **early engagement** with the different groups and organisations that would be involved in establishing the clinical trials and/or participating in them. Throughout the process, **persistence**, **openness**, and **willingness** to speak with many stakeholders was key. We also ensured there was trust, respect, and transparency within our team.

Case Study 3 (continued)

Approach 1: Networking and collaboration

Starting clinical trials for new therapies and incorporating these therapies into routine clinical care was complex and required networking and collaboration with a wide range of people and organisations, including families with SMA, regulatory agencies, clinical teams, and decision makers.

Other approaches, which happened at the same time as we were establishing our centre as a clinical trial site, included:

- Learning more about SMA and the people we hoped to treat to better understand the numbers of people affected, their individual and family characteristics, and the natural course and effects of SMA
- **Listening to the lived experience and unmet needs of families with SMA and clinical teams**, which ensured our goals were person-centred and focused on the patient and their families or carers
- **Engaging and collaborating** with many different people and organisations. This includes research ethics and governance committees, pharmacists, clinical teams, advocacy and community stakeholders, Government agencies, scientific communities, clinical experts, and companies in the biotechnology/pharmaceutical industry
- Taking part in regular meetings and workshops to update collaborators and share learnings and education
- Applying existing care guidelines and models of care with the help of our **networks**
- Speaking to and **partnering with experts** to help us understand and complete regulatory processes that would allow us to start the clinical trials
- **Working with behavioural scientists, health economists, and data experts** to fill evidence gaps
- **Collaborating with international networks** to utilise their expertise.

Case Study 3 (continued)

After the clinical trials were complete, we wanted as many children as possible to be able to receive the new treatment. However, clinical trials have strict inclusion and exclusion criteria – meaning entry to a trial may be limited based on age, type of SMA, and how affected the child is. We had limited evidence that the treatments worked in children who did not meet the inclusion and exclusion criteria for our trials, so we collated real-world data about how children responded to treatment outside of a clinical trial. Capturing this real-world evidence was critical to expanding access to the treatment.

Approach 4: Patience, persistence and flexibility

To expand treatment access to more children, the team established a patient registry to generate 'real world' data, a challenging but necessary task.

Sharing knowledge, clinical evidence, and lived experience

We looked for opportunities to share knowledge and education, increase awareness, collaborate, and secure funding. We were **constantly communicating and educating people**, including developing resources. We presented a unified clinical expert voice and utilised the Australian Neuromuscular Network **community of practice** around the clinical centres so that we were all consistent in care and practice and learning as a group. We also **engaged with the media** when appropriate.

Approach 2: Communication and storytelling

Communicating an expert and evidence-based message was a critical part of SCHN's approach.

They also ensured communication from all partners was consistent.

To better understand the needs of families, the team listened to people's personal stories.

Case Study 3 (continued)

Having clear evidence of better health outcomes and stories reflecting lived experience was a powerful combination for setting advocacy goals. **The patient voice and lived experience** was also critical for setting our priorities.

Persistent communication to convey consistent, accurate and cohesive messages and goals

As a community we established three priorities for consistent communications:

- Access to therapies
- Early diagnosis (through newborn screening)
- Reproductive screening.

Many different approaches were used to communicate these key messages and influence decision makers, including:

- Planning and aligning messaging
- Having **multiple conversations** with decision makers
- Developing **case studies describing the benefits to individual patients** to help decision makers clearly see the value of the treatment
- Ensuring that when decision makers spoke with families and advocacy organisations, they heard the **same message** they had heard from clinicians and researchers
- Writing briefs for executive leadership; this alone was usually not able to bring about the outcomes we were seeking, so we had to support these briefs with **further communication and discussions**.

Key messages

This case study highlights the value of:

- **Networking and collaboration** with a wide range of people and organisations to gain diverse input
- **Listening to and sharing the voices** of people living with a rare disease
- **Consistent messaging and a unified voice** from clinical experts and people living with a rare disease
- **Patience and persistence** to continue even when the first attempt was not successful

"When decision makers spoke with families and advocacy organisations, they heard the same message they had heard from clinicians and researchers."

Case Study 4

Partnering with university researchers to fill gaps in knowledge and resources about *SATB2* Associated Syndrome

SATB2 Associated Syndrome is a rare neurodevelopmental condition caused by changes in the *SATB2* gene.[21-23] Individuals with *SATB2* Associated Syndrome have intellectual disability, delays in developing motor skills such as sitting and walking, speech difficulties or absent speech, behavioural challenges, low muscle tone, and distinct facial features. Many organ systems may be involved, including the heart, teeth, bones and spine, and gastrointestinal system.

Our advocacy goal

SATB2 Connect is a not-for-profit organisation that supports families and their loved ones diagnosed with *SATB2* Associated Syndrome in Australia, New Zealand, and neighbouring countries in the Asia Pacific region.[24] Our goal was to develop a paper outlining the gaps and unmet needs of individuals and families affected by *SATB2* Associated Syndrome. The aim of the paper was to inform a research plan for our community, with our community.

Our first step was to seek advice and support from researchers at four universities and **build a network**, which we formalised as our Research Advisory Committee.

Approach 1: Networking and collaboration

Establishing early partnerships with four different universities led to the development of a Research Advisory Committee, which informed the research and priorities.

The researchers were interested in working with patient-led organisations to improve the quality of life of people diagnosed with *SATB2* Associated Syndrome and their families. They offered to co-supervise students with our organisation to **co-design** and conduct much needed research **together** – our families are the experts and the co-designers of the research.

Approach 3: Co-design

Working closely with researchers and people with lived experience helped focus the research and ensure it was relevant to the *SATB2* community.

Case Study 4 (continued)

Through surveys and one-on-one interviews, the students have been collating data about gaps and challenges in health care, education, support and National Disability Insurance Scheme (NDIS) funding, therapy, accommodation, and supportive workplaces.

Defining the project through collaboration and co-design

We came to our partner universities with our **community's needs and experiences** and then together, identified, and prioritised the areas of research we needed to focus our efforts on. Our families have been respected as lived experience experts and are **co-designers** of the research.

Throughout the project, we reconvened and discussed who the research audience would be, and what the content and target journals should be for the research, being **flexible and yet persisting** in reaching our goals.

Approach 4:
Patience, persistence and flexibility

The project required a considerable time commitment from the SATB2 Connect team, with patience and persistence required to see the project to completion.

Forming partnerships through storytelling and collective advocacy

We have achieved the most success by **sharing our family experiences**. We keep the accounts simple and related to the target audience's individual interests. It is helpful to learn about the individuals and organisations we are pitching our ideas to before meeting with them.

We were most successful when we approached universities as an established organisation with a **clear message summarising our mission and goals**. We had less success when approaching universities individually.

Case Study 4 (continued)

Approach 2:
Communication and storytelling

A clear message summarising missions and goals was critical to gain support and buy-in from potential research partners.

The power of connection and being part of a wider community

We **made use of our connections** with national and international patient organisations, medical, health, and research networks. We understand our community is small and that's why it's important to **be a part of the wider rare disease community** and to advocate broadly for chronic and complex health conditions.

Networking with other like-minded organisations and those who share many of the common challenges of *SATB2* Associated Syndrome helped with identifying the right decision makers and best advocacy channels. Events and conferences run by peak bodies such as RVA and other organisations gave us more opportunities to learn, share and network.

Our **international network** helped us grow our understanding of what our community needs and how we can work together and make the most of our resources and time.

Investing in the future

We believe involving university students in our work builds their capacity and expertise, and hopefully ignites a passion in them to continue to be involved in, or even lead, future rare disease research.

Some of the challenges faced by small organisations like ours

- Knowing where to start, how to start and when to start
- Finding research opportunities at no cost to the rare disease organisation
- Gaining knowledge and an understanding of the best language to communicate with academia, the rare disease sector, and our community
- Knowing how to best engage our community
- Keeping students participating in group research engaged and interested
- Having enough time available to achieve all our goals
- Knowing how to keep the research scope reasonable to avoid researcher burnout.

Case Study 4 (continued)

Key messages

This case study highlights the value of:

- **Networking and collaboration** to build a Research Advisory Committee
- **Lived experience and storytelling** to engage collaborators and help them understand the needs and priorities of individuals and families
- **Co-designing** projects with the rare disease and scientific community to take advantage of lived and scientific expertise
- **Patience, persistence, and flexibility** in re-defining goals and planning new approaches when initial strategies (for example, meeting potential partners as individuals) were not successful

"Although the tasks ahead are challenging for patient organisation leaders, we are often reminded that we are the driving force for change; we are the parents, caregivers and families of those individuals living with chronically complex medical challenges – this is why our passion is further ignited for positive outcomes."

Section 3

Approaches and tools to support effective advocacy

Why is advocacy important in rare disease?

Change benefitting Australians living with a rare disease is often driven by effective advocacy undertaken in response to issues, needs or gaps. There is great complexity and unmet need in rare disease, and Australia's health and other systems are often not fit for purpose for rare disease.

> Even though our organisation is small, fairly new and runs completely with volunteering members, we have strived to access opportunities because we see a need and many gaps in many areas of our systems."
>
> **Rare disease organisation leader**

Getting involved in advocacy

In the case studies about Action Plan development and the legalisation of mitochondrial donation, both RVA and the Mito Foundation drove positive change by co-ordinating and supporting collective advocacy to the Australian Government. However, effective advocacy can happen in many other ways.

Most people in rare disease advocacy become involved through need rather than choice. Advocacy is often driven by recognising a gap or unmet need and then exploring ways to address these gaps. Examples include:

- A parent seeing a promising clinical trial overseas and advocating for a site in Australia
- A person seeing that treatments for their condition are available overseas and advocating for access to this treatment for themselves and their rare disease community
- A clinician wanting to improve clinical care and have access to the latest diagnostics
- A person wanting to speak up about their rights to obtain appropriate, timely, and accessible health care, in line with the Australian Charter of Healthcare Rights
- A health professional, passionate about delivering high quality holistic care, wanting to write a submission for an enquiry into accessible disability services.

Section 3 (continued)

Potential challenges in rare disease advocacy

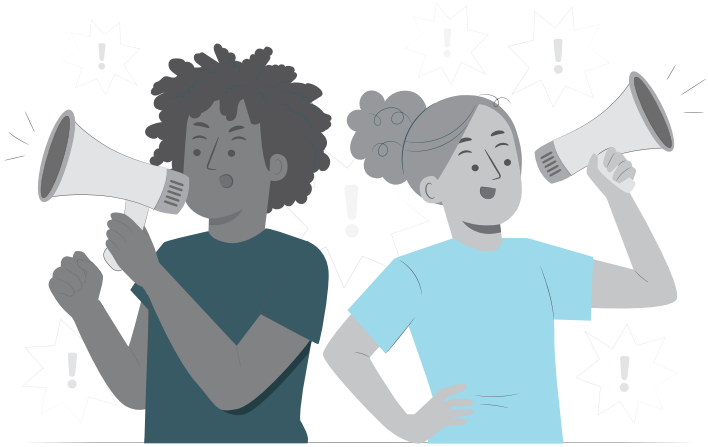
Living with a rare disease requires resilience, persistence, and optimism, as does rare disease advocacy.

The following challenges are relevant for all rare disease advocates across the rare disease sector:

- Lack of data/evidence
- Lack of knowledge and awareness
- Lack of resources
- The need to learn ‘on-the-go’
- Health and support systems that are complex and fragmented
- Small numbers of people with the rare disease.

> Every vulnerable community gets tired of advocating and asking to be respected, but really, the responsibility needs to start to shift toward the sector to understand why that respect is not there in the first place.”
>
> **Person living with an undiagnosed rare disease**

Rare Voices Australia’s Engaged, Ethical and Effective: A Guide For Rare Disease Organisation Leaders in Australia is a useful tool for rare disease groups and organisations.[25] It contains resources and checklists, including advice on advocacy and community engagement.



Section 3 (continued)

Advocating for changes to Government policy

Government policy is the basic agreed principles by which government is guided.[26] Government policies can have a significant impact on the rare disease sector, for example, by determining the funding and operation of the health services caring for people living with a rare disease.

The Action Plan is an Australian Government **policy framework** and the key policy framework for rare disease in Australia. Generally, a policy framework provides an overarching structure that guides how policies and procedures will be developed, approved, communicated and reviewed.[27] Policy frameworks also communicate a course of action for a particular issue or group of people.

Aligning advocacy for changes in Government policy with the Action Plan is criticial to ensure effective implementation.

A wide range of Australian Government policies relate to rare disease. These include:

- Health technology assessments
- Medical research funding
- Data generation and management
- Genomic testing
- Newborn screening
- Hiring and training the health workforce
- Management of chronic conditions
- Hospital funding
- Disability
- Education and employment.

To advocate effectively, it is important to first understand these different policy areas, the relevant legislation, policies and processes, and key decision makers. As Australia has a **federated health system**, understanding the different responsibilities of each level of government (state and federal) is essential for targeted and effective advocacy.

The Australian health system is made up of both public and private service providers. All levels of government, non-government organisations, health insurers, and individuals (via philanthropic funding) can fund health service research, design, delivery, and evaluation.

More information about the Australian health system can be found in the Toolkit companion document *How the health system works in Australia*.

Section 3 (continued)

Tips and strategies for effective advocacy

As the national peak body for Australians living with a rare disease, RVA offers guidance to groups/organisations on effective rare disease advocacy. RVA acknowledges that there are many different advocacy styles and approaches.[25]

As most rare disease advocacy is likely to be ongoing and have multiple strategic and long-term goals, RVA recommends placing strong relationships at the centre of any advocacy approach. Although public aggressive approaches that ambush people may result in quick wins, they may jeopardise future advocacy.

Robust discussions, which are sometimes necessary, are always best engaged in privately and in an appropriate manner. Fostering trust and credibility with key stakeholders and decision makers are key to securing meetings.

RVA has several useful tools to help groups/organisations get started on their advocacy journey. More resources to help with advocacy are highlighted at the bottom of this section.

Rare Voices Australia's recommended advocacy style[25]

- Polite persistence as a starting point
- Relationship-based
- Robust conversations, when needed, held privately
- Solutions-focused and pragmatic
- Evidence-based
- Collaborative and dependent on building and maintaining relationships
- Credible and independent
- Make time to understand and leverage unique positioning
- Person-centred with a multistakeholder approach
- A balance of long- and short-term approaches

Section 3 (continued)

Advocacy Self-Evaluation. This was written for groups/organisations but could be adapted for individual advocacy or the work of informal alliances.

PRINT

Identify your advocacy strengths and development areas using this self-evaluation tool.
Tick each box that applies to your organisation.

- ☐ Advocacy is aligned with the strategic objectives of the organisation.
- ☐ Advocacy reflects the priorities of the community.
- ☐ Advocacy is informed by a comprehensive understanding of the policies, frameworks and decision makers that influence key advocacy issues.
- ☐ Evidence and data to support advocacy positions has been collected from reputable sources with the relevant expertise (for example, scientific and medical advisory committees, clinicians).
- ☐ Other organisations that are advocating on similar issues have been identified, including opportunities to work together.
- ☐ Consideration has been given to the advocacy style that is most appropriate for the organisation's goals (for example, long term, relationship-based or single issue, quick wins).
- ☐ Advocacy is informed by the values and ethical principles of the organisation.

Reproduced from Engaged, Ethical and Effective: A Guide For Rare Disease Organisation Leaders in Australia with permission from RVA.[25]

Section 3 (continued)

Tips and strategies to help ensure advocacy is strategic, targeted, and credible

PRINT

Have a clear objective/s

- What is/are the problems or issues?
- What action or resolution are you seeking?

Be informed

- Who can influence this issue?
- Are there policies or legislation that are relevant?
- What evidence demonstrates the problem and supports the proposed solutions?
- Are there others with similar interests I should work with?
- What are some potential objections or barriers to my request?

Identify a suitable advocacy style

- Authentic styles, aligned with your organisation's strategic intent and values that take into account the complexity of the issue and the number of stakeholders you may need to influence, will be most effective

Follow up

- Repeatedly, persistently, politely

Reproduced from Engaged, Ethical and Effective: A Guide For Rare Disease Organisation Leaders in Australia with permission from RVA.[25]

Section 3 (continued)

Aligning advocacy with the National Strategic Action Plan for Rare Diseases

It is helpful to develop an advocacy strategy that aligns with the messaging and language in the Action Plan. This helps the rare disease sector to communicate a consistent and unified message. RVA's resource Amplifying Advocacy Using the National Strategic Action Plan for Rare Diseases provides information, case studies and examples to show how the Action Plan can be used for advocacy.

The template on the following page is an excerpt from this guide and provides examples of specific advocacy activities that can be supported by using the Action Plan.

Further reading

Tools and resources to advocate for policy change across the globe as well as regionally, nationally and locally (Rare Diseases International). A collection of tools and resources to support advocacy at all levels.

Share4Rare toolkit for patient advocacy (Share4Rare). A practical resource to help plan, build, test, communicate and launch a new advocacy effort.

View from inside: Patient advocacy in guiding policy development for metabolic disorders (Journal of Inherited Metabolic Disorders). This article by Nicole Millis, RVA's Chief Executive Officer, offers valuable insights into advocacy and offers ideas and suggestions.

Why effective advocacy is more important than ever (LinkedIn). This blog post by Nicole Millis provides valuable advice on advocacy and what effective advocacy looks like.

Section 3 (continued)

Ways to use the Action Plan in advocacy activities	Upcoming activity
Submissions into upcoming inquiries and Government consultations	Example: submission lodged for the Parliamentary Inquiry into approval processes for new drugs and novel medical technologies in Australia
When writing to Members of Parliament at State and Federal level	Example: write to my local Member of Parliament to request a meeting to discuss my community's rare disease
When writing funding/sponsorship submissions, project proposals, and policy submissions	Example: RVA referring to the Action Plan when requesting support from the RVA Round Table of Companies
Communicating with your community about your organisation's goals and priorities	Example: media releases, social media posters or e-newsletters
When providing consumer comments for health technology assessment processes	Example: submitting comments to PBAC consumer comments

Reproduced from Amplifying Advocacy Using the National Strategic Action Plan for Rare Diseases with permission from RVA.[28]
PBAC, Pharmaceutical Benefits Advisory Committee; RVA, Rare Voices Australia.

Section 3 (continued)

Approaches highlighted in the Toolkit case studies

Approach 1. Networking and collaboration: How to find your allies and experts

This approach includes the following key tools:

1. The World Bank's Community of Practice Toolkit
2. Rare Voices Australia's Policy page
3. Partnership/collaboration checklist
4. Considerations when planning collaborations with people living with a rare disease
5. Involve Australia's Guidelines for Community Involvement in Genomic Research
6. Medicines Australia's Working Together Guide
7. Steps to evaluate and improve collaboration

What is networking and collaboration, and why is it so important in rare disease advocacy?

As rare diseases are so rare, people living with a rare disease may feel isolated and alone. While there are many different rare diseases, they share countless commonalities. The Action Plan highlights that because of the unique challenges of living with a rare disease, individuals and organisations in the rare disease sector need to work together.

Types of collaborations and partnerships in the rare disease sector

There are many types of collaborations and partnerships.

In a **formal partnership** between two or more organisations, there is shared understanding and typically a written agreement among the organisations making the roles and responsibilities of each organisation clear. For example, RVA partners with over 100 rare disease-specific organisations.

A **community of practice** is a group of people who come together regularly to work towards common goals. These are particularly helpful in rare disease, where knowledge may be scarce. A shared forum can help fill gaps and solve problems that cannot be resolved alone. For example, the RArEST Australian Rare Disease Organisation Community of Practice (ARDO-CoP) and the Rare Disease Project ECHO® Clinical Community of Learning Practice, formed as part of the RArEST Project.

Section 3 (continued)

> If you've got other people around you that are trying to do similar things ... there will be opportunities for us to continue to work together on specific issues. And that's really powerful.
>
> **RArEST Australian Rare Disease Organisation Community of Practice member**

Networks and alliances across organisations and/or countries are particularly important for rare diseases, since each rare disease typically has a small number of patients, families, and experts. For example, Rare Diseases International's Global Network for Rare Diseases and Genomics in the Community.

The World Bank's Community of Practice Toolkit provides guidance and resources for setting up and running community of practice.

Panels, boards, and working groups can be established to address specific challenges, share expertise, or guide policy and practice for rare diseases. Clinicians may be invited to join boards to provide clinical expertise, and individuals may be asked to provide lived experience and personal insights into the priorities of people living with a rare disease.

Formal consultations are usually initiated by governments, regulatory bodies, or international organisations. In a formal consultation, a range of individuals and organisations are asked to provide their input.

The Policy page on RVA's website provides several sample submissions for government consultations.

Research partnerships are partnerships between groups conducting research into rare diseases and one or more individuals or organisations within the rare disease sector, for example, a peak body, rare disease organisation, university, research institute, hospital, health care network, or biotechnology company.

Section 3 (continued)

RVA's Research Partnership Guidelines show how RVA ensures that research partnerships are aligned with the policies and goals of their organisation. Other organisations may consider developing similar guidelines and processes.

Research partnerships can be initiated by rare disease organisations

SATB2 Connect established research partnerships with four different universities to drive further research into *SATB2* Associated Syndrome.

"Our first steps ... were to seek advice and support from researchers at four universities and build a network which we formalised as our Research Advisory Committee."

SATB2 Connect

Many partnerships and collaborations are **cross-sectoral**, that is, they bring together individuals and organisations from different parts of society.

For example, a collaboration may be between the government, health services, or non-government organisations, including rare disease groups/organisations. Cross-sectoral or multi-sectoral collaborations aim to incorporate a wide range of experiences, opinions and expertise into policies, services, and resources.

Cross-sectoral partnerships

The formal collaborations formed between the Australian Government, RVA, their partner groups/organisations and people living with a rare disease helped shape the Action Plan and ensure the priorities of all groups were heard and incorporated.

"Working in partnership with Australians living with a rare disease was key and helped identify the goals and actions of the Action Plan."

Rare Voices Australia

Section 3 (continued)

How to seek out and establish collaborations and partnerships

PRINT

When considering starting a partnership or collaboration, the following checklist may be helpful.

Partnership and collaboration checklist

- ☐ Does your potential partner's vision and priorities align with those outlined in the Action Plan?
- ☐ Does their mission align with yours?
- ☐ What aspect of rare diseases do they address? (for example, clinical, policy, other advocacy)
- ☐ What expertise are they offering?
- ☐ Is the collaborator part of your organisation or another one?
- ☐ Are there any risks?
- ☐ Are there any conflicts of interest? If so, how will these conflicts of interest be managed?

Collaborating with people with lived experience

Including people with lived experience of a rare disease and their voices in all aspects of health care, including policy development, research, system design, the development of education materials, and care planning, has numerous benefits.[29] These include ensuring health care is person-centred and efficient, resources are applicable and useful, and research is aligned with the needs of the rare disease community.

Rare disease groups and organisations often work beyond capacity and are staffed by volunteers who may be living with a rare disease themselves or caring for someone who has a rare disease. It is recommended that people's capacity, health literacy, accessibility needs, and practical constraints are considered when asking for input.

Networking to unify and amplify the voice of lived experience

In each of the four case studies, partnerships with people living with a rare disease helped inform the goals and priorities of the advocacy.

Section 3 (continued)

Ways people living with a rare disease are involved

People living with a rare disease and rare disease groups/ organisations are often involved in:

- Leading or chairing a committee or working party
- Presenting training or education
- Designing or conducting research projects
- Developing and reviewing, strategy, research, or resources
- Selection panels and grant review panels
- Focus groups or interviews.

It is important to be aware of and remove any barriers to participation for people living with a rare disease and their families/carers. This will involve discussion with each individual, but typically would include considerations such as making the timing of meetings family-friendly, ensuring interactions with other stakeholders feel welcoming and safe, ensuring the meeting spaces are accessible, or holding meetings virtually so travel is not required. Consumer remuneration may be a potential way to remove barriers. For example, Health Consumers NSW suggest appropriate rates according to the level of consumer engagement or involvement.

PRINT

Considerations when planning collaborations with people living with a rare disease

- ☐ Are the people living with a rare disease representing their whole community, or just their individual experience?
- ☐ Is it better to seek a diverse range of individual perspectives, or work with representatives from organisations who already represent a wide range of individuals?
- ☐ If working with individuals, how diverse is the group and how can you support diverse representation?
- ☐ Are there specific access needs for those with communication differences and how can those be addressed?
- ☐ If working with groups/organisations, how do they seek and reflect the perspectives of those they represent, including priority populations?
- ☐ Is there any additional training or support that may be helpful for the individual or group and how could that be provided?

Section 3 (continued)

Involve Australia's Guidelines for Community Involvement in Genomic Research provides genomic researchers in Australia with practical information for involving community members effectively and meaningfully in research projects.

It is important to include all groups in the rare disease community. This includes priority populations such as:

- People with a disability
- Aboriginal and Torres Strait Islander people
- People from culturally and linguistically diverse backgrounds
- People living with an undiagnosed rare disease
- People experiencing socio-economic disadvantage
- People from regional, rural, and remote regions.

It may not always be possible to directly engage with members of all priority populations. In this case, consider collaborating with the peak bodies representing these communities. This is particularly important for Aboriginal and Torres Strait Islander peoples, and culturally and linguistically diverse communities, where cultural safety is critical. Peak bodies and health advocacy organisations with broad representation include the Consumers Health Forum of Australia, the Australian Multicultural Health Collaborative, the National Rural Health Alliance, National Aboriginal Community Controlled Health Organisation (NACCHO) and the Federation of Ethnic Communities' Councils of Australia (FECCA).

The RVA Education webinar Developing the Rare Disease Resources Collections for Priority Populations provides insights into developing resources for people living with a rare disease from priority populations, including working with community and peak bodies who have established relationships and trust with priority populations.

To learn more about collaborating with people with lived experience, see Approach 3: Co-design.

Medicines Australia's Working Together Guide provides guidance on how the pharmaceutical and biotechnology industry can work with consumer groups.

Section 3 (continued)

Collaborating with scientific and medical experts

All advocacy benefits from the involvement of credible and expert scientific and medical advisers.[25] Scientific and medical advisers can be involved in many different ways. Examples include; helping to ensure information and educational resources are accurate and factual, assisting with submissions, or gaining the support of their colleagues and institutions to drive change in their own settings.

The value of working together

Collaborating isn't just about building relationships – it helps to get things done.

"With many people and groups involved they each contribute a few pieces of the puzzle – it can't be done alone."

Clinician, SCHN

Collaborating with the pharmaceutical and biotechnology industry

Many pharmaceutical and biotechnology companies fund basic and clinical research into rare diseases. They may also fund and support programs for people living with a rare disease, educational programs for health care professionals, or projects to build health literacy among people living with a rare disease.

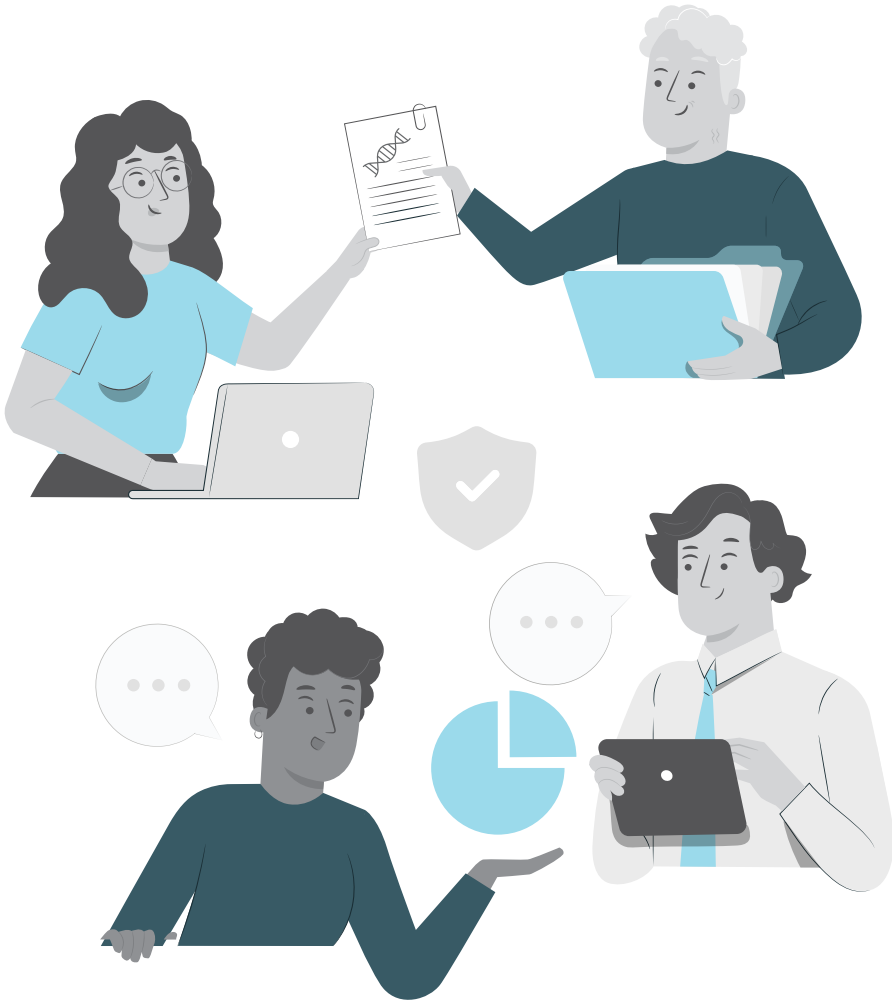
Working with these companies can provide much-needed financial support, however, it is important to ensure that priorities align, and there is mutual benefit for all involved. Chapter 6 in RVA's Engaged, Ethical and Effective: A Guide for Rare Disease Organisations provides additional insights and advice on grant funding and sponsorship from the pharmaceutical and biotechnology industry.

Section 3 (continued)

A valuable and productive collaboration between a rare disease group/organisation and a pharmaceutical/biotechnology company requires a strong relationship that is based on core principles that include:[30]

1. Respect for independence
2. Achieving and maintaining public trust
3. Open communication
4. Confidentiality
5. Accountability
6. Successful collaborations that focus on the health consumer and their carers
7. Management of conflicts of interest.

The steps outlined in the tool on the next page may help with evaluating and improving collaborations.[31]



Regular communication is important

It keeps networks connected and maintains relationships.

"[we brought] together stakeholders regularly to update and share learnings and education."

Clinician, SCHN

Section 3 (continued)

Steps to evaluate and improve collaboration

PRINT

STEP 1 – Find a common purpose

- ☐ Partners have compared their respective organisation's goals and advocacy objectives
- ☐ All partners are aware of their part to play

STEP 2 – Increase collaborative literacy

- ☐ Partners have a shared understanding that collaboration is vital to the success of their work and that each partner has equally valuable expertise to offer

STEP 3 – Knowing who is 'in the tent'

- ☐ Partners know who their partners are and why they have those particular partners

STEP 4 – Monitor development

- ☐ Partners monitor the extent to which their partnership has formed, developed, and transformed over time

STEP 5 – Assess the strength of the partnership

- ☐ Partners are aware of the current and potential future strength of the partnership
- ☐ Decisions are made about continuing the strength of the partnership or how to strengthen the partnership

STEP 6 – Assess collaboration on an ongoing basis

- ☐ Partners regularly assess if others are still invested in, and working towards, a common purpose
- ☐ The quality of the collaborations, for example decision-making processes and ensuring partnerships are equal, authentic, and productive, has been considered

Adapted from the Handbook on Planning, Evaluating, and Improving Collaboration for Oral Health Programs.[31]

Section 3 (continued)

Approach 2. Communication and storytelling: How to create clear messaging and share personal stories safely

This approach includes the following key tools:

1. Living with a rare disease: Digital mental health resources
2. Checklists for listeners and storytellers

What is effective communication?

Just as there is no one way to advocate, there is no one way to communicate. There are many different communication styles and approaches. Effective communication is crucial in advocacy as it empowers people to convey their messaging in a way that positively influences decision makers.

Once you have a clear idea of which stakeholders you are targeting, ensure your messaging speaks to the intended audience. This is so they can grasp the key points quickly and participate in the call to action if they choose to.

Clear, accurate, consistent, and evidence-based information is especially important in rare disease. It builds credibility and reduces the likelihood of confusion, overwhelm and misinterpretation.

> We are people, there is the sector and the dollars, there's diagnosis and there's research. There are all these things, but it really often forgets... the human experience."
>
> **Person living with an undiagnosed rare disease**

Quality communication is essential for building and maintaining relationships/partnerships, maintaining attention, fostering connection, and enhancing engagement and participation. It can also help to secure the resources required to work towards a shared goal.

Section 3 (continued)

Typically, it's best to map out a communications plan and test what is and isn't working through whatever measures are available, for example; social media analytics, or email open rates. It is important to be open to learning and receiving feedback while continuing to advocate.

Keep in mind that 'communication' is a broad term and encompasses a range of approaches. This could include social media, traditional media, email marketing, follow-up phone calls and meetings. Different approaches may require different language and messaging. For example, communicating via LinkedIn generally requires professional language and is useful for professional networking. In contrast, Facebook is more suited to communicating with families and communities.

"When there is so much that needs to be done, it can be tempting to call for a whole list of changes, resulting in a myriad of messaging that can be overwhelming, confusing, or unfortunately, come across as venting or complaining. A clear, targeted, and nuanced message is often much more effective."

Nicole Millis[32]

Storytelling

A powerful way to communicate is through **storytelling**. The experiences of people living with a rare disease can be valuable in several ways.

These may be incorporated into effective systemic advocacy as part of a wider strategy:

- To demonstrate key messages in advocacy or policy documents, emphasising the power of co-design with people living with rare diseases
- To support submissions to the Federal or State Governments for funding research and/or new technologies
- To provide insights into the impact and challenges of living with rare disease. These insights can increase the impact of educational resources and guidelines

Storytelling is not only an approach that can be used effectively by people living with a rare disease. Health professionals may also have powerful experiences to share, which can highlight the needs of people living with a rare disease. They can also identify and call out the barriers and gaps in the health system and suggest solutions that may improve care and support.

Section 3 (continued)

> Stories of someone's diagnostic journey and experience managing their condition could be beneficial to clinicians and other health professionals to identify symptoms, make a prompt diagnosis and optimise treatment."
>
> **Person living with a rare disease**

Key considerations

While storytelling is a powerful approach, there are several things to consider to ensure storytelling is carried out safely and effectively.

- The person sharing their story must consent to their story being shared, and should know where, how and for what purpose their story will be shared. For example, Rare Voices Australia has an online consent form for their Share Your Story section.
- Every person is an individual, so their story and the way they share their story will be personal and require individual approaches.
- Sharing stories can be distressing. Sharing RVA's digital mental health resources fact sheet with the storyteller may be helpful.
- Some people with a rare disease are asked to share their story repeatedly. It is important to consider how many times someone has shared their story. When the same people share their story frequently, it can re-traumatise them. The checklists on pages 48-49 may be helpful for people who are sharing their stories and for those listening to and sharing other people's stories.

Section 3 (continued)

> Lived experience is very lengthy and I think that story telling is a series of experiences. It can be traumatic to revisit issues. It is also bold to share such life details and a person's willingness to do this must be valued."
>
> **Person living with a rare disease**

The [Living with a rare disease. Digital mental health resources] fact sheet, developed by the RArEST Project, provides mental health and wellbeing support for people living with rare disease. It is a useful resource to share with storytellers. It includes links, resources with useful information, support, and skills.

The checklists were adapted from the Health Issues Centre's [The Kit: A guide to the advocacy we choose to do][33] and [Rare Disease 101 Australia, Lesson 4: Respectful and Effective Communication].[34] **Please note:** these checklists are generalised and should be used in conjunction with culturally safe practices for Aboriginal and Torres Strait Islander peoples, and culturally and linguistically diverse communities.

Section 3 (continued)

Checklist for listeners

PRINT

When listening to someone's story:

- ☐ Clarify the purpose of their sharing. Do you have consent to share their story?
- ☐ Be mindful of the space in which they are sharing, and create a safe and non-judgmental space
- ☐ Show you are actively listening, by making eye contact and being aware of their body language
- ☐ Engage respectfully, giving them the time they need to share, and allowing them to stop whenever they need
- ☐ Refrain from interrupting the person sharing their story
- ☐ Be mindful of your own biases and reserve personal opinions and judgement
- ☐ Explore nuances, whilst being mindful to not press for matters that are sensitive or traumatising
- ☐ Ensure the stroryteller, and others in the group (if relevant) are aware of support services should they be traumatised by information shared

Section 3 (continued)

Checklist for storytellers

PRINT

When sharing your story:

- ☐ Remember, your story is yours to tell and you are not obligated to share
- ☐ Decide on what you are comfortable sharing, and what you need to make the process easy
- ☐ Prepare, as much as possible, thinking about what you would like to say and identifying topics that are ‘off limits’
- ☐ Consider the ‘take home message’ you want to leave with your audience. What would you like them to do in response?
- ☐ If telling your story in first person (I, me) feels too personal, consider using third person (he/she/they)
- ☐ Consider the sharing environment you are in and the purpose of sharing your story
- ☐ Ensure the audience listening to your story has asked for your consent
- ☐ Ensure the audience you are sharing your story with has been transparet about the use of your story and willing to maintain your privacy if you wish

Section 3 (continued)

Supporting the storyteller

It is important to support the storyteller, as sharing a story can be emotional and potentially distressing. Everyone has the right to share their story in a way that feels safe and comfortable.

Ways to provide support include:

- Making sure the storyteller is prepared before they start sharing their story
- Having a trained and experienced staff member present when stories are being shared
- Establishing guidelines for what is appropriate to share
- Encouraging storytellers to share their stories in ways that are strengths-based and solutions-focused
- Checking in with the storyteller afterwards.

> Engagement needs to be with both individuals and organisations, to have more of a breadth and diversity in those voices, also being mindful of fatigue. Depending on the population, the same people are being tapped on the shoulder all the time."
>
> **Person living with a rare disease**

Those who are newly diagnosed or early in their rare disease journey may need close monitoring and/or additional support. These people may feel traumatised or demotivated by the stories of people further along in their rare disease journey.

Information about mental health support resources is available on the Rare Awareness Rare Education (RARE) Portal. State-specific resources can be found under the 'mental health' subheading of each respective state and territory.

People who are distressed during or after the storytelling process can be referred to crisis help lines:

Lifeline: **13 11 14**

Beyond Blue: **1300 22 4636**

13 YARN: **13 92 76**
(Aboriginal and Torres Strait Islander service)

Section 3 (continued)

Approach 3.
Co-design: How to apply co-design to ensure the needs and priorities of all rare disease stakeholders are included

This approach includes the following key tools:

1. Questions to ask before starting a co-design project
2. Organisational readiness assessment
3. NSW Health Agency for Clinical Innovation's Co-design toolkit
4. Australian Healthcare and Hospitals Association's Experience based Co-design Toolkit
5. Reflection questions for after a project is complete

What is co-design?

Co-design is a process and approach that involves all stakeholders working as equals in leadership, design, delivery, and decision-making. Essentially, it is the creation of **solutions *by* and *for* people impacted by those decisions**. This is especially important in rare disease where there is much unmet need and limited evidence-based solutions. Co-design is different from consultation, because co-designers have an active role in **contributing to the process on an ongoing basis**.

It is important to note:

- Differences of opinion will occur – these differences should be seen as learning opportunities and points of collaboration, not a barrier to progress.
- There should be opportunities for people living with a rare disease to take on active roles in line with their capacity and willingness to do so.

There are several phases of co-design, as described in Table 2, on the following page.

Section 3 (continued)

Table 2. Co-design phases

PRINT

Design phase

- Identify the issue or common goal
- Understand the gaps and opportunities
- Be aware of where different expertise lies
- Establish a relationship with the community

Delivery phase

- Convene your co-designers
- Be patient and open to the back-and-forth (iterative) nature of co-design
- Gather expertise, especially lived experience, via interviews, written communication, surveys or other methods preferred by the community
- Do not expect everyone to agree

Evaluation phase

- Sense-check that the desired outcome was achieved
- Determine if the outcome is appropriate and aligns with the needs of the community
- Evaluate and review the methodology for appropriateness

Section 3 (continued)

Co-design requires careful thought and planning. The tool below may be helpful before starting a co-design project.

Questions to ask before starting a co-design project

PRINT

- ☐ How will I go about ensuring the rare disease community is involved in my work from the very beginning, including identifying needs and goals?
- ☐ How will I go about seeking expertise and input from the rare disease community? What questions will I ask?
- ☐ Who will I engage with from the rare disease community? How do I plan to engage with them?
- ☐ What steps do I need to take to achieve the goal or vision in partnership with the rare disease community?
- ☐ Is our process iterative and does it bring along the rare disease community with every iteration?
- ☐ How can I establish a safe space for storytelling? What measures do I need to take and what are the key considerations?
- ☐ What are the risks for my storytellers in sharing their lived experience? How will I support storytellers sharing their lived experience and set up mechanisms for follow-up?
- ☐ Which other key stakeholders from the sector do I need to align with?
- ☐ How can we frame and strengthen our message as a collective, and ensure the rare disease community are part of crafting that message?

Section 3 (continued)

Getting ready to co-design

Co-design may be more challenging and complex for larger organisations, especially those that have policies and procedures that do not historically include consultation and co-design.

Completing the checklist on the next page before starting a project may help organisations assess whether they are ready for co-design, and if not, where there are opportunities to strengthen for co-design. This checklist on page 55 was adapted for the rare disease sector from the NSW Health Agency for Clinical Innovation's Organisational co-design readiness assessment.[35]

The challenge of co-design

"We find true co-design very difficult to achieve. We do involve the mito community [those with lived experience of mitochondrial disease] at all stages of our work and we're getting better at working in partnership."

Mito Foundation

Section 3 (continued)

PRINT

Is your organisation ready to co-design	Strongly agree	Agree	Unsure	Disagree	Strongly disagree
Co-design is central to our organisation and is supported at all levels.					
Our approach to co-design is iterative - priorities, focus and tasks can change or evolve.					
We are aware of the unique challenges of living with a rare disease and value the expertise people living with a rare disease offer.					
We do, or are open to, co-designing with everyone in the rare disease sector, understanding that solutions to complex issues require many people or groups.					
We involve or plan to involve people living with a rare disease in the design, delivery and evaluation of policies, services, medications, or therapies that impact them.					
When planning our co-design approach, we budget for remuneration, and consider the timing needed for storytelling.					
When we plan to co-design with the rare disease community (including rare disease organisations), we are aware of, and sensitive to, the challenges of living with a rare disease and make adjustments accordingly.					
When we co-design or plan with the rare disease community, we authentically and meaningfully consider those in the community from Aboriginal and Torres Strait Islander backgrounds, culturally and linguistically diverse communities, and other priority populations. We ensure relevant expertise is sought to help us engage.					
We have appropriate mechanisms and/or a staff member to support people before, during or after sharing their story, and for those listening to stories.					

Section 3 (continued)

Co-design tools

Many tools have been developed to help guide co-design. Most are not specific to rare disease and may need adapting to consider the unique challenges faced by the rare disease community.

NSW Health Agency for Clinical Innovation's Co-design toolkit provides guidance and resources to help health services adopt a co-design approach, including suggestions for working together in co-design.

The Australian Healthcare and Hospitals Association's Experienced Based Co-design Toolkit helps bring the Australian health sector together in authentic and equal partnership for co-design.

After a co-design project is complete

The questions below may be helpful when a co-design project is complete.

Reflection questions for after a project is complete PRINT

- ☐ Did I successfully involve people living with a rare disease in my work? How could I do this better in the future?
- ☐ Did I effectively embed the lived experience of people living with a rare disease in my work? How do I know this? Did I evaluate my work?
- ☐ Did I support people living with a rare disease and did I provide a safe place for sharing their lived experience?
- ☐ What did I learn from this process that I did not know before?
- ☐ Is our process iterative and does it bring along the rare disease community with every iteration?
- ☐ Did anything work exceptionally well that I would do again? Are there certain things that I would do differently and why?

Section 3 (continued)

Further reading

A Guide to Build Co-design Capability (Agency for Clinical Innovation). This resource helps to build capacity of health services to co-design with people with lived experience of a health condition.

Ways of Working Together in Co-design (Agency for Clinical Innovation). An interactive toolkit that provides details about co-designing with others in the health sector.

Spectrum of Public Participation (International Association of Public Participation). A visual tool to help organisations understand the degree of community or individual participation.

Consumer participation strategies (Centre for Culture, Ethnicity and Health). A visual tool that depicts the many ways a community or individual can participate. It is a culturally and linguistically diverse-specific tool.

Co-production guidelines (UNSW Disability Innovation Institute). Recordings of webinars and helpful guidelines in accessible and user-friendly formats.

Co-design with Aboriginal and Torres Strait Islander peoples and communities (Metro North Health Queensland Government). Tips and helpful resources for co-designing with Aboriginal and Torres Strait Islander peoples.

Consumer and Community Involvement Program e-Course (CCI Program). A shared community of people with lived experience, universities, health and medical research institutes, health services, and non-Government organisations.

Section 3 (continued)

Approach 4. Patience, persistence, and flexibility: How to keep going despite the uncertainty and challenges – rare disease challenges are complex and often require complex solutions

This approach includes the following key tool:

1. Tips for staying on track and beating burnout

Advocacy can be hard work and may become overwhelming. There will be challenges and setbacks, and success may not come easily or quickly. A good starting point is to break the goal down into smaller steps.

> I learnt pretty early on that effective advocacy is much harder and more time-consuming than it looks."[32]
>
> **Nicole Millis, CEO, Rare Voices Australia**

The case studies in this Toolkit show common characteristics of organisations that have achieved positive outcomes:

- They are **patient**.
  For example, the Mito Foundation's campaign to change the law took several years. The Action Plan was not written overnight. It takes time to listen to all voices, formulate a plan, test if that plan might work, and then re-assess if it will not.
- They are **persistent**. It is very unlikely that advocacy efforts will be successful on the first attempt.
  For example, SATB2 Connect approached multiple potential partners, and continued to engage different organisations even after their initial attempts were unsuccessful.
- They are **flexible**. The path to success is not always linear, and following the same steps as others will not always work. Being willing to re-assess and revise initial planning after a setback is critical.
  For example, SCHN realised that further communication and discussion with decision makers was needed after initial briefs did not bring about the desired outcomes.

Section 3 (continued)

> Key approaches were patient and persistent relationship-building through meetings with key political supporters... and knowing when to go hard and when to be patient with the process."
>
> **Mito Foundation**

> Throughout the project, we reconvened and discussed who the research audience would be, and what the content and target journals should be for the research, being flexible and yet persisting in reaching our goals."
>
> **SATB2 Connect**

Continuing... even when the going gets tough

It can be disheartening when the best efforts of the team do not achieve the desired outcomes. There may be individual or collective burnout – when people lose motivation and energy to keep trying. The tool below provides strategies to keep the momentum going and combat burnout.

Tips for staying on track and beating burnout PRINT

- ☐ Celebrate the small wins
- ☐ Enlist new team members to bring more energy and/or ideas
- ☐ Acknowledge that it is okay not to have all the answers
- ☐ Seek out and listen to the ideas of others – especially when problems arise and a new approach is needed
- ☐ Practice gratitude to yourself, those you work with and even those who have challenged you (since they are helping you to grow and learn)
- ☐ Prioritise self-care, especially if you are leading and/or carrying a lot of responsibility

Glossary

Term	Definition
Advocacy	Sharing views and opinions with others to create positive change
Advocate	Publicly support or speak up for a cause
Centre of Expertise	Provides specialist care for people with specific conditions or groups of conditions (for example, neuromuscular conditions or other groups of rare diseases)
Chronic	Long-term
Co-design	A process and approach that involves all stakeholders working as equals in leadership, design, delivery, and decisionmaking
Cross-sectoral	Across more than one sector of the community or society e.g. primary care and specialist care; health and education
Multi-sectoral	Across multiple sectors of the community or society
Policy framework	An overarching structure that guides how policies and procedures will be developed, approved, communicated and reviewed
Rare disease sector	People living with a rare disease, their families and carers, and the health professionals, organisations and systems that care for them
Stakeholder	A person, group or organisation that has an interest in decisions or activities that are important to them
Systemic change	Change that affects the systems involved in providing care and support to people living with a rare disease

Abbreviations

Abbreviation	Definition
ARDO-CoP	Australian Rare Disease Organisation Community of Practice
NDIS	National Disability Insurance Scheme
NHMRC	National Health and Medical Research Council
PBAC	Pharmaceutical Benefits Advisory Committee
RArEST	Rare Disease Awareness, Education, Support and Training
RVA	Rare Voices Australia
SCHN	Sydney Children's Hospitals Network
SMA	Spinal muscular atrophy
UNSW	University of NSW

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