



Australia's Top 10 Rare Disease Research Priorities

The Kids Research Institute Australia and Rare Voices Australia

June 2025

Acknowledgement of Country

We acknowledge Aboriginal and Torres Strait Islander people as the Traditional Owners of Country throughout Australia and as a priority population of the rare disease sector. We also pay our respects to Elders – past and present – and generations of Aboriginal and Torres Strait Islander people now and into the future.

Acknowledgements

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Ethics Approval Statement

Ethical approval was obtained for this project from The University of Western Australia Human Research Ethics Committee (2024/ET000158).

Funding Statement

This project was funded by RVA. The outcomes of this work are based on views gathered from individuals across the rare disease sector and do not necessarily represent the views of RVA or the Project Executive Team.

About Rare Voices Australia

Rare Voices Australia (RVA) is the national peak body for Australians living with a rare disease. RVA provides a strong, unified voice to advocate for policy as well as health, disability and other systems that work for people living with a rare disease. RVA's person-centred focus sees them working with all key stakeholders, including Australians living with a rare disease, governments, key peak bodies, researchers, clinicians and industry. RVA's work is non-disease specific and is based on the commonalities of rare disease. RVA advocates for the best outcomes for Australians living with a rare disease and focuses on rare disease policy, processes and systems.

In 2018, the Australian Government commissioned RVA to lead the collaborative development of the National Strategic Action Plan for Rare Diseases¹ (the Action Plan). The Action Plan is the first nationally coordinated effort to address rare diseases in Australia and was informed by an extensive multistakeholder consultation process led by RVA. The Action Plan was launched in February 2020, with bipartisan support. Since its launch, RVA has continued working alongside the rare disease sector to lead the collaborative implementation of the Action Plan.

[Rare Voices Australia website](#)

About The Kids Research Institute Australia

The Kids Research Institute Australia (The Kids), formerly Telethon Kids Institute, is an independent, not-for-profit medical research institute committed to improving the health and wellbeing of children through excellence in research. As one of the largest and most successful medical research institutes in Australia, The Kids brings together researchers, community, medical professionals, policy makers and funders who share a vision to improve the health and wellbeing of children through excellence in research.

[The Kids Research Institute Australia website](#)

About the Method

To establish Australia's Top 10 Rare Disease Research Priorities (the Top 10 priorities), this project followed a modified version of the James Lind Alliance Priority Setting Partnership methodology. This modified approach was first used at The University of Western Australia and The Kids in 2016.² A former James Lind Alliance Co-Chair advised on the development of the modified model used in this project.

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Executive Summary

Limited data, knowledge and treatment options are common in rare disease. These factors contribute to significant unmet need experienced by the estimated 2 million Australians living with a rare disease.^{1,3} This equates to 8 percent of Australians.^{1,3} Most Australians living with a rare disease experience high levels of symptom complexity and ongoing health and psychosocial challenges. These long-term daily impacts meet the Australian Government's definition of a disability⁴, impacting all facets of life.

Launched in 2020, the Australian Government's National Strategic Action Plan for Rare Diseases¹ (the Action Plan) is the first nationally coordinated effort to address rare diseases. The Action Plan highlights that research into rare diseases must inform evidence-based policy across all systems, including the health, disability, social/welfare, mental health, education, employment and housing systems.³

Australians living with a rare disease are best placed to articulate the widespread impacts of rare disease and the priorities for rare disease research in Australia. In line with RVA's person-centred approach, the project captured the voices of 185 Australians living with a rare disease, their families, carers, health professionals and community representatives, to identify Australia's Top 10 Rare Disease Research Priorities (pictured below).

About This Work

Research and Data is one of three main Pillars in the Action Plan, which highlights a lack of research into rare diseases despite recognised gaps in knowledge.

This project was driven by Action Plan Priority 3.2, "*Develop a national research strategy for rare disease to foster, support and drive all types of research for rare diseases, contributing to agreed priorities and systematically addressing gaps.*" To progress this priority, RVA, led by its Scientific and Medical Advisory Committee, engaged The Kids to conduct a Priority Setting Partnership project, based on a modified James Lind Alliance Priority Setting Partnership methodology. The aim was to establish Australia's Top 10 Rare Disease Research Priorities based on what matters most to people living with a rare disease, including their parents, carers, health professionals, and other rare disease community representatives.

Australia's Top 10 priorities for the future of rare disease research are intended to guide the strategic efforts of the rare disease sector, including RVA as the national peak body, researchers, and health and medical research funders.

Australia's Top 10 Rare Disease Research Priorities

1

Development of, and access to, the best treatments and cures, including clinical trials



2

Awareness and education for health professionals and service providers



3

Diagnosis, including screening



4

Networked centres of expertise



5

Data collection and use



6

Genetic testing



7

Person-centred best practice care and support



8

Psychosocial and mental health impacts and support



9

Access to resources and information for Australians with rare disease, families and carers



10

Disease progression and, where relevant, prevention measures



Introduction

Collectively common, rare diseases affect approximately 2 million Australians.^{1,3} Common challenges include difficulty obtaining an accurate diagnosis, a lack of treatment options and inadequate, fragmented care. Rare diseases, like many other chronic diseases, are often serious and progressive. They typically display a high level of symptom complexity and are a significant cause of ongoing health, psychosocial challenges and disability.

Rare disease research is not only critical to closing knowledge gaps, but also an integral part of best practice clinical care to improve the health and wellbeing outcomes of Australians living with a rare disease. Despite broad recognition of the need for rare disease research, significant gaps remain. Gaps include limited rare disease knowledge and treatments, significant diagnostic delays and misdiagnoses, and high levels of unmet need. These gaps are evident across various government systems from health and disability to housing and education.

This report presents the outcomes of a rare disease Priority Setting Partnership (PSP) project – a person-centred approach to identifying and prioritising domains of research that matter most to people living with a rare disease, and those who care for and support them. By directly involving key stakeholders in shaping Australia's rare disease research agenda, this PSP honours people's lived experience. It also supports broader national policy directions, including Australia's commitments to equity, consumer involvement in health and medical research, and the increasing focus of health and medical research funders on areas of unmet need.

Rationale and Aim

As the number of known rare diseases grows rapidly, knowledge gaps widen. Ongoing research is required to fill these gaps and ensure the health and wellbeing needs of Australians living with rare diseases are met. However, available data and knowledge in rare diseases is limited in comparison to more common diseases, so it is difficult to know where to focus efforts and allocate resources. For many rare diseases, there are several barriers to effective research, and no active research programs. One of the biggest challenges is the misperception that research into diseases with inherently small patient numbers has a negligible impact.

Depending on the specific rare disease, research priorities can be different. For example, while funding for translational research may be important for many rare diseases, some rare diseases are not yet in the position to prioritise translational research. For certain rare diseases, the unmet research needs are basic discovery research or investment into data collection and natural history studies. Exemplified by the Action Plan, the rare disease sector has adapted to move beyond these challenges by working collaboratively on commonalities and progressing evidence-based systemic reforms that crosscut rare diseases and systems.

Guided by RVA's Scientific and Medical Advisory Committee, this project aimed to progress Priority 3.2 of the Action Plan, *“Develop a national research strategy for rare diseases to foster, support and drive all types of research for rare diseases, contributing to agreed priorities and systematically addressing gaps”*.

‘Person-centred’ is a foundation principle of the Action Plan. In line with this foundation principle, this project set out to understand the most important gaps and unanswered questions regarding rare disease research according to Australians living with rare disease, their families/carers, health professionals and advocates. This person-centred approach further aligns with Australian Government policy and practice, including the National Health and Medical Research Council's efforts to value the lived experience of consumers and community organisations in health and medical research. It also aligns with the co-design approach of the National Health and Medical Research Strategy development team.

Methodology

This project followed a modified James Lind Alliance (JLA) Priority Setting Partnership (PSP) methodology. The JLA PSP methodology was originally developed to identify and prioritise health research questions relating to a specific condition or healthcare setting that are important to patients, their families and clinicians. A list of the top research priorities, often presented as a ‘Top 10’, is collaboratively developed, reflecting the needs and views of end-users.⁵ The modified version of the JLA PSP method was first developed, and used, in Western Australia in 2016.² This methodology has been successfully used to identify priorities for long-term conditions including cancer,⁶ chronic pain⁷ and asthma.⁸ Throughout this project, adherence to the modified JLA PSP method was guided by an expert advisor. Additional advice was intermittently sought from a former JLA PSP facilitator, based in the United Kingdom.

Figure 1 shows the 5 stages of the modified JLA PSP methodology followed for this work. A detailed description of the methodology is presented in Appendix B.

Figure 1.

Modified JLA PSP Methodology Followed.

Modified JLA PSP Methodology



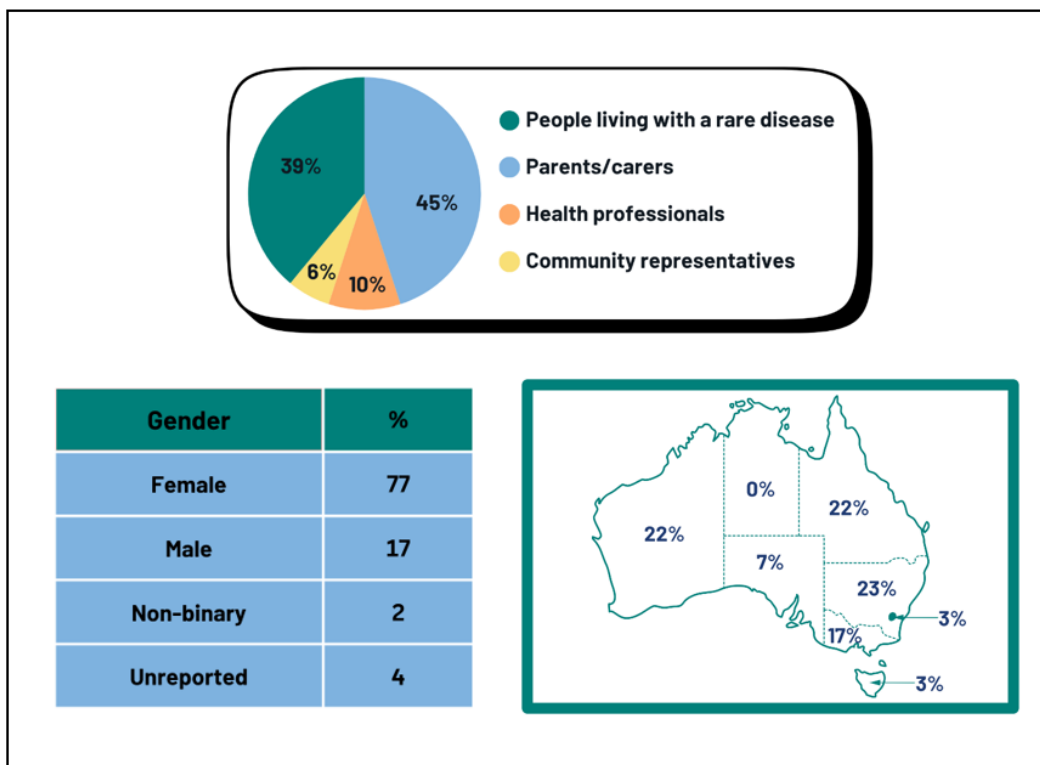
Detailed Findings

19 Themes Emerging from the Survey

185 people completed the survey, including individuals living with rare disease, their families/carers, health professionals and community representatives (Figure 2). From the 585 free text comments/statements/questions contributed, 19 themes and associated subthemes were identified (see Table 3 in Appendix B). These themes were developed with rare disease subject matter experts and in line with language used in the Action Plan.

Figure 2.

Survey Respondent Demographic Data



The frequencies (number of responses) of each theme presented in Table 1 are for informational purposes only. In keeping with the modified JLA PSP methodology followed, frequencies were not provided to consensus workshop attendees to avoid influencing theme discussions at the workshops.

Table 1. 19 Themes from Survey Responses, Presented in Order of Frequency

Theme	Frequency of Theme in Survey Comments/ Statements/Questions
Treatments and cures	80
Person-centred best practice care and support	57
Awareness and education for health professionals and service providers	51
Disease progression and, where relevant, prevention measures	45
Diagnosis, including screening	43
Impacts of rare disease	38
Data collection and use	30
Management of symptoms and comorbidities	25
Access to resources and information for Australians living with rare diseases, their families and carers	19
Psychosocial and mental health impacts and support	19
Networked centres of expertise	18
Support for parents and carers	15
Access to the best available treatments, including clinical trials	14
Quality of life	14
Genetic testing	13
International collaboration	11
Disability guidance and support	9
Support and information for reproductive choice	9
Care and support for all Australians, including priority populations	6

Differences and Common Ground: Comparing the Top 10 Rare Disease Research Priorities from Community and Health Professional Perspectives

Consensus workshops play a critical role in this methodology, allowing for deeper discussion and the consideration of nuances and perspectives that may be difficult to express through a survey alone. As a result, the final order of priorities differs from the initial survey rankings. This reflects a more informed and collaborative understanding shaped by meaningful dialogue. Rare disease community members and health professionals attended separate consensus workshops. Participants were asked to decide which of the 19 themes, presented in Table 1, should be included in Australia's Top 10 Rare Disease Research Priorities. Figure 3 presents the similarities and differences in the Top 10 priorities between these two groups.

Figure 3.

Top 10 Rare Disease Research Priorities from Consensus Workshops



The overwhelming majority of themes chosen as part of the Top 10 priorities overlapped between community and health professionals. These similarities are highlighted in yellow

in Figure 3. By contrast, the white boxes in Figure 3 draw attention to notable differences in prioritised themes between community and health professionals.

When discussing the themes, consensus workshop attendees were asked to consider whether any themes could be combined or if any were missing. In both the community and health professional consensus workshops, it was agreed the following two themes should be combined:

- Access the best available treatments, including clinical trials; and
- Treatments and cures

Community consensus workshop attendees agreed on the following wording for this combined theme:

‘Development of and access to the best treatments and cures, including clinical trials’

By contrast, health professionals agreed to the following wording:

‘Treatments and cures, including clinical trials’

The project executive team decided the phrasing of the combined theme would remain consistent with what was agreed to in the community consensus workshop, as (1) this consensus workshop had a greater number of participants, and (2) the title was more descriptive and better upheld all aspects of the original theme *‘Access to the best available treatments, including clinical trials’*, and associated subthemes.

Participants in the community consensus workshop also agreed to move the theme *‘Support and information for reproductive choice’* to become a subtheme of *‘Genetic testing’*. The project executive team decided to keep this change in the Top 10 priorities, not only because it honoured the strong collective opinion of those at the workshop, but also because it broadened the inclusivity of the original theme *‘Genetic testing’*, allowing for richer interpretation and broader application of the research priority.

Aligning Perspectives: Australia’s Top 10 Rare Disease Research Priorities

To achieve a Top 10 list, ranked themes from consensus workshops were averaged. This section details the Top 10 priorities, and underlying subthemes, for future rare disease research prioritisation in Australia (Table 2).

Table 2. Australia’s Top 10 Rare Disease Research Priorities and Underlying Subthemes.

Priority	Subthemes
<p>Priority 1. Development of, and access to, the best available treatments and cures, including clinical trials</p> <p><i>“How can we develop therapies that not only manage symptoms but also target the underlying causes of rare diseases?”</i></p>	<ul style="list-style-type: none">• Gene and cell therapy and emerging technology expansion• Funding for new treatments• Repurposing treatments for rare diseases• Timely drug development and access, including faster approval processes for reimbursed treatments• Identify biomarkers to support development of rare disease treatments, including for personalised medicine• Improve availability of clinical trials for rare disease treatments in Australia• Improve timely and equitable access to clinical trials for Australians living with rare diseases
<p>Priority 2. Awareness and education for health professionals and service providers</p> <p><i>“Overall practitioners need to understand... that often the patient knows more about the condition and that can be burdensome ... It’s important to listen to what works for them.”</i></p>	<ul style="list-style-type: none">• Improve knowledge and awareness of rare diseases, including for diagnosis, referral pathways and treatments• Reduce stigma and the need for patient/carer advocacy• GP education• Encourage critical, ‘outside the box’ thinking

Priority	Subthemes
	<ul style="list-style-type: none"> • Improve health professional collaboration and knowledge sharing
<p>Priority 3. Diagnosis, including screening</p> <p><i>“Long diagnostic pathways negatively impact families in so many ways, including limiting eligibility for treatments, care, and support services.”</i></p>	<ul style="list-style-type: none"> • Clearer diagnostic pathways • Reduce misdiagnoses • Improve access to screening • Clearer pathways for timely and appropriate access to rare disease specialists and knowledge, both locally and interstate in the pre-diagnostic stage • Develop/improve diagnostic criteria and rare disease classifications • Support for individuals and families in the pre-diagnostic stage
<p>Priority 4. Networked centres of expertise</p> <p><i>“There needs to be a coordinated model of care and support for families to navigate care and services, receive mental health assistance, and access expert health care and clinical trials.”</i></p>	<ul style="list-style-type: none"> • Improve visibility and access to rare disease specialists • Improve access to care for adults living with a rare disease • Centralise access to rare disease information and research • Integrate peer connection opportunities
<p>Priority 5. Data collection and use</p> <p><i>“I think there is not much data available on rare disease from a research perspective which would underpin finding a treatment/improve care.”</i></p>	<ul style="list-style-type: none"> • National statistics, including prevalence data • Shared databases and registries • Frameworks for data sharing, both nationally and internationally • Standardised patient reported outcome measures/metrics for rare disease
<p>Priority 6. Genetic testing</p> <p><i>“More focus on those who are carriers to help them with family planning and support.”</i></p>	<ul style="list-style-type: none"> • Improve accessibility and awareness of genetic testing and pre-conception testing

Priority	Subthemes
<p><i>“Better and faster access to genetic testing.”</i></p>	<ul style="list-style-type: none"> • Develop and improve genetic testing protocols • Genetic testing for family members • Support and information for reproductive choice
<p>Priority 7. Person-centred best practice care and support</p> <p><i>“What good care and support look like and how to implement these in our complex systems (health, disability, education, social care etc) ... Rare disease impacts the whole family.”</i></p>	<ul style="list-style-type: none"> • Optimise health services • Equitable access to care • Identify and implement models of coordinated and integrated care • Transition to adult care • Family-centred, holistic care models
<p>Priority 8. Psychosocial and mental health impacts and support</p> <p><i>“What is best practice for supporting the psychological and emotional needs of people diagnosed with a rare disease?”</i></p>	<ul style="list-style-type: none"> • Identify psychosocial and mental health impacts of living with rare diseases, including the impact of a rare disease diagnosis on family members • Improve access to tailored psychosocial and mental health support for individuals and families • Develop psychosocial support services and resources tailored to rare disease
<p>Priority 9. Access to resources and information for Australians with rare disease, families and carers</p> <p><i>“Very little has ever been explained to me regarding my child's diagnosis and prognosis.”</i></p>	<ul style="list-style-type: none"> • Identify gaps in rare disease information, research and data to inform development of information and resources • Maintain updated and relevant information sources on rare diseases • Improve awareness of, and access to, available resources and supports for individuals and families • Develop plain language summaries of research and information on rare diseases
<p>Priority 10. Disease progression and, where relevant, prevention measures</p>	<ul style="list-style-type: none"> • Identify rare disease causes

Priority	Subthemes
<i>“Why do some individuals with the same rare disease have vastly different experiences in terms of severity and progression?”</i>	<ul style="list-style-type: none">• Understand environmental and lifestyle influences• Improve understanding of variations in symptoms and outcomes

Limitations, Challenges and Learnings

Sampling

- 77% of respondents to the survey and 85% of consensus workshop attendees identified as female
- Only 1.6% of survey respondents identified as Aboriginal or Torres Strait Islander
- Underrepresentation of bereaved families

Unique Challenges/Barriers to Participation

People living with a rare disease, their families and carers face unique challenges, including managing complex health needs, fatigue and care responsibilities. These factors can significantly limit their ability to participate in research. While efforts were made to ensure communication and recruitment materials were developed in simple English for translation and to be accessible to people with different abilities, the lack of easy read formats and translation of materials into other languages may have been a barrier to participation for some individuals.

While the JLA PSP methodology offers a powerful, person-centred approach, it is time-intensive and involves multiple stages of engagement—requirements often not feasible for many in the rare disease sector. This includes health professionals who, in some cases, are the sole clinicians in Australia with specialised rare disease expertise. These barriers make it difficult to engage large numbers of participants. Despite these barriers, the level of participation in this study exceeded expectations, underscoring the value and urgency of this work within the rare disease sector.

Learnings

Significant multifaceted gaps in data and knowledge, and high levels of unmet need, uniquely inherent to rare disease, are overwhelming for the entire rare disease sector. This includes policymakers, clinicians, researchers, industry and most profoundly, individuals living with rare disease and their families. Consequently, asking stakeholders to prioritise many nuanced, complex and equally critical issues, especially under time constraints in a workshop setting, is inherently difficult. This tension was reflected in feedback shared by some workshop attendees.

Concluding Remarks

The Importance and Application of Australia's Top 10 Rare Disease Research Priorities

Australia's Top 10 Rare Disease Research Priorities provide an important foundation to inform and guide future rare disease research efforts. Progress across the Top 10 priorities will assist in the ongoing collaborative implementation of the Australian Government's National Strategic Action Plan for Rare Diseases (the Action Plan).

Drawing on the lived experience of Australians living with rare diseases, their families, health professionals and other advocates, the Top 10 priorities reinforce national and global efforts to ensure research is inclusive, person-centred and responsive to areas of highest unmet need.

Beyond addressing persistent knowledge gaps, rare disease research is critical to best practice clinical care and plays a vital role in improving the health and wellbeing outcomes of Australians living with a rare disease. Nonetheless, despite clear and pressing needs, rare disease research remains significantly underfunded in Australia.

The Top 10 priorities present a timely opportunity to attract greater attention and investment from health and medical research funders, research institutions, policymakers and industry. They are a valuable resource for all rare disease stakeholders, including researchers, clinicians, health service planners, advocacy organisations and government. The Top 10 can be used with confidence as they reflect the lived experiences and expertise of individuals and families affected by rare disease, and the health professionals who support them. Importantly, all 19 themes and subthemes identified in this work, regardless of their inclusion in the Top 10 priorities, collectively contribute to a robust and person-centred research agenda that acknowledges the full breadth of the rare disease experience.

Finally, in addition to driving national efforts, this work contributes to a growing international movement that recognises the value of shared research priorities. The Top 10 priorities can potentially inform international collaborations, guide global research funding, and align with key policy frameworks, including those championed by the International Rare Diseases Research Consortium (IRDiRC) and the United Nations Resolution on Addressing the Challenges of Persons Living with a Rare Disease.

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Appendices

A. Governance

Project Advisory Group and Steering Committee

A Project Advisory Group and Steering Committee informed each step of the project.

The Project Advisory Group met on four occasions over the duration of the project. Members played a key role in guiding the project and providing perspectives and expertise on important issues that may impact their community's involvement in the research. They supported recruitment of participants, and provided input into project methods, survey design and dissemination of survey and recruitment materials.

The Project Advisory Group comprised of:

- Three people with lived experience of rare diseases
- Two parents/carers of people living with a rare disease
- Two clinicians working with people living with rare diseases
- One representative from RVA
- One researcher involved in rare disease research
- One expert advisor with extensive experience using the modified JLA PSP process

The Steering Committee met on three occasions over the duration of the project, and provided strategic direction, advice and oversight, ensuring the aims and outcomes of the project were achieved. They also ensured the community voice remained integral to the project.

The Steering Committee comprised of:

- Two members of RVA's Scientific and Medical Advisory Committee
- Two representatives from RVA
- One organisational representative with expertise in health research and policy
- One person with lived experience of rare diseases
- Two members of the research team
- One expert advisor with extensive experience using the modified JLA PSP process

B. Detailed Methodology

Recruitment

Participants were recruited to the study via RVA's social media and email databases, as well as via Project Advisory Group and Steering Committee networks. Flyers were circulated through these recruitment channels with a link for people interested to provide informed consent and participate in the study. Upon completion of the online survey, participants who indicated interest in attending a consensus workshop were prompted to provide their contact information via a separate link. This ensured the identity of survey respondents remained anonymous. When recruiting attendees for the online consensus workshops, individuals who provided their contact information were contacted directly to register their interest.

Stage 1: Online Survey

An online survey was conducted to elicit respondents' questions relating to rare diseases that they would like answered by research. Survey questions are detailed below.

Survey questions for people living with rare diseases, their families/carers and community representatives

1. What research do you think is most needed in rare disease?
[Free text]
2. What questions, issues or gaps do you think we should understand about rare disease?
[Free text]

Survey question for rare disease health professionals

1. What are the most important unanswered questions about the impacts of rare diseases on the people you care for, or things you think we need to know or better understand about rare diseases?
[Free text]

Demographic data, including gender, age, and residing state, were also collected as part of the survey (See Figure 2).

Stage 2: Content Analysis

All free-text survey responses were analysed by the research team and rare disease subject matter experts. The analysis was an iterative process, beginning with data familiarisation and initial theme generation from 585 text comments. This was followed by refinement and reorganisation of the 19 research themes and the development of subthemes, which

provided further context and detail (See table 3). The Project Advisor provided a review of the final themes from a non-researcher perspective to ensure themes and subthemes were simple and representative of the survey responses. Input was also sought from the Project Advisory Group and Steering Committee to ensure the phrasing of themes was clear and appropriate.

Stage 3: Evidence Checking

A total of 19 themes generated during Stage 2 were assessed against available evidence in the literature to determine the extent to which they may have been addressed or answered by existing research. Two members of the research team reviewed existing systematic reviews using Medline, Embase, CINAHL, and the Cochrane Database of Systematic Reviews to assess the level of evidence relating to each theme.

Themes were considered unaddressed if: (1) no up-to-date systematic review on the topic had been published; (2) published systematic reviews indicated insufficient evidence; (3) available evidence was not considered high quality; or (4) the available evidence was inconclusive. Any themes that were addressed were to be reviewed by the Steering Committee, and if recommended, removed from the list of themes to be discussed at the consensus workshops. None of the 19 themes satisfied the criteria for being comprehensively addressed by existing systematic reviews. Therefore, all themes were considered relevant gaps and were taken to the consensus workshops for prioritisation.

Stage 4: Consensus Workshops

Online consensus workshops were held to identify the Top 10 rare disease research priorities from the 19 themes identified in Stage 2. Separate consensus workshops were held for health professionals and community members (people living with or caring for people living with rare disease and community advocates/representatives). The rationale for this was to enable open conversations.

A single 3-hour consensus workshop was held for health professionals, whilst the community consensus workshop was divided into two sessions of approximately 90-minutes each. These two sessions were held one-week apart to accommodate accessibility requirements for this group.

Participants received pre-reading materials to review prior to the consensus workshop, including an agenda, a document outlining what to expect during the consensus workshop, and a list of the themes and subthemes generated from the online survey responses. Themes were shared with participants in alphabetical order to avoid unintentional bias. Each theme was attributed a letter identifier (from 'A' through to 'S') to enable easier referencing to themes during the consensus workshop discussions.

Consensus workshops were led by two facilitators with experience in the modified JLA PSP methodology, and small groups were facilitated by members of the research team and representatives from RVA. Both consensus workshops followed a similar format, detailed below, which involved a combination of whole group and small group discussions to reach consensus on the Top 10 priorities.

Consensus Workshop Part 1: Theme Discussion

In the first small group discussion, each participant was asked to reflect on the themes to identify their top and bottom three themes. They were also asked to consider whether any themes could be combined or if any themes were missing. Following this, each facilitator reported their small group discussion back to the larger group to reach consensus on themes that could be combined or added. Consensus was achieved for each discussion point when a majority vote was received.

Consensus Workshop Part 2: Theme Ranking/Prioritisation

In the second small group discussion, the group facilitators supported participants to rank themes from most to least important. The rankings assigned by each group were used as scores and averaged to determine the combined ranking of all themes and arrive at the Top 10 priorities. At the end of each consensus workshop participants worked collectively to discuss and vote on the final Top 10 rare disease research priorities.

Workshop Evaluation

At the conclusion of consensus workshops, participants were asked to complete an online evaluation survey to provide feedback on the process. Of the 34 attendees across both workshops, 21 completed the evaluation survey.

Over half of participants reported that the consensus workshop(s) either completely or mostly met their expectations (57%), with an additional 19% reporting they did to ‘a fair amount’. The vast majority felt that their questions were answered adequately (86%) and that they had sufficient opportunity to put forward their ideas and priorities for research (81%). Just over two-thirds of participants felt that the presentations provided enough information (71%) and the priority setting process was explained sufficiently (71%).

Participants reported that the best thing about the workshop(s) was:

- *“The diversity of attendees and their connection to rare disease - which made the conversation well rounded”*
- *“A mixed group of passionate people who want to make a difference”*
- *“Being able to share and collaborate on research priorities with other people across Australia”*

- *“Being involved in such an important process”*
- *“Collaboration and hearing other viewpoints, seeking input from the community on rare disease priorities”*

Participants reported that the worst thing about the workshop(s) was:

- *“Not enough time”*
- *“Given huge variation in conditions some topics did not lend themselves to being combined or ranked higher for some than others. The time restriction was difficult in some ways but with more time I’m not sure a consensus would ever be arrived at”*
- *“I felt it was really rushed. Too rush[ed] to feel sure we even made the correct choices. I think it was a large and difficult task and should have been done as 2 half day sessions or a full day, I also don't think it was clearly stated these were research priorities. There's a big difference in asking people what they think are important issues and rank them, versus what are actual research topics that should potentially be funded. I think this needed explaining and regularly reminding, like what the purpose was. People seemed more saying what's important to them.”*
- *“No collaboration between clinicians and patients/carers”*
- *“Trying to hear others opinion over Zoom and make sure everyone was having a say”*

Stage 5: Top 10 Rare Disease Research Priorities, Including Ranking of Remaining Themes

Results from consensus workshops with health professionals and community were combined to arrive at the final Australia’s Top 10 Rare Disease Research Priorities. To do this, the rankings of themes at the end of both workshops were averaged to achieve an overarching Top 10 list. Table 3 shows the combined ranked list of themes and subthemes.

Table 3. Combined Final Rankings of all 19 Themes Following the Consensus Workshops.

Rank	Theme
1*	Access to the best available treatments, including clinical trials <ul style="list-style-type: none">• Improve availability of clinical trials for rare disease treatments in Australia• Improve timely and equitable access to clinical trials for Australians living with rare diseases
1*	Treatments and cures <ul style="list-style-type: none">• Gene and cell therapy and emerging technology expansion• Funding for new treatments• Repurposing treatments for rare diseases• Timely drug development and access, including faster approval processes for reimbursed treatments• Identify biomarkers to support development of rare disease treatments, including for personalised medicine
2	Awareness and education for health professionals and service providers <ul style="list-style-type: none">• Improve knowledge and awareness of rare diseases, including for diagnosis, referral pathways and treatments• Reduce stigma and the need for patient/carer advocacy• GP education• Encourage critical, ‘outside the box’ thinking• Improve health professional collaboration and knowledge sharing
3	Diagnosis, including screening <ul style="list-style-type: none">• Clearer diagnostic pathways• Reduce misdiagnoses• Improve access to screening• Clearer pathways for timely and appropriate access to rare disease specialists and knowledge, both locally and interstate in the pre-diagnostic stage• Develop/improve diagnostic criteria and rare disease classifications• Support for individuals and families in the pre-diagnostic stage
4	Networked centres of expertise <ul style="list-style-type: none">• Improve visibility and access to rare disease specialists• Improve access to care for adults living with a rare disease• Centralise access to rare disease information and research• Integrate peer connection opportunities

Rank	Theme
5	<p>Data collection and use</p> <ul style="list-style-type: none"> National statistics, including prevalence data Shared databases and registries Frameworks for data sharing, both nationally and internationally Standardised patient reported outcome measures/metrics for rare disease
6^	<p>Genetic testing</p> <ul style="list-style-type: none"> Improve accessibility and awareness of genetic testing and pre-conception testing Develop and improve genetic testing protocols Genetic testing for family members
6^	<p>Support and information for reproductive choice</p> <ul style="list-style-type: none"> Provide better education for individuals and families who are carriers of a genetic rare disease Increase available supports for individuals and families who are carriers of a genetic rare disease, including better access to genetic counselling
7	<p>Person-centred best practice care and support</p> <ul style="list-style-type: none"> Optimise health services Equitable access to care Identify and implement models of coordinated and integrated care Transition to adult care Family-centred, holistic care models
8	<p>Psychosocial and mental health impacts and support</p> <ul style="list-style-type: none"> Identify psychosocial and mental health impacts of living with rare diseases, including the impact of a rare disease diagnosis on family members Improve access to tailored psychosocial and mental health support for individuals and families Develop psychosocial support services and resources tailored to rare disease
9	<p>Access to resources and information for Australians with rare disease, families and carers</p> <ul style="list-style-type: none"> Identify gaps in rare disease information, research and data to inform development of information and resources Maintain updated and relevant information sources on rare diseases

Rank	Theme
	<ul style="list-style-type: none"> • Improve awareness of, and access to, available resources and supports for individuals and families • Develop plain language summaries of research and information on rare diseases
10	Disease progression and, where relevant, prevention measures <ul style="list-style-type: none"> • Identify rare disease causes • Understand environmental and lifestyle influences • Improve understanding of variations in symptoms and outcomes
11	Impacts of rare disease <ul style="list-style-type: none"> • On individuals • On families and carers • On education and employment • On health sector and wider community • Financial hardship and societal costs
12	International collaboration <ul style="list-style-type: none"> • Increase collaboration between Australian and international rare disease health professionals • Increase rare disease data-sharing between Australia and other countries
13	Management of symptoms and comorbidities <ul style="list-style-type: none"> • Develop strategies to improve management of rare disease symptoms and reduce their severity and impact • Improve understanding and management of comorbidities • Investigate exercise and dietary supplements for condition management
14	Support for parents and carers <ul style="list-style-type: none"> • Self-advocacy support and tools • Identify what forms of support parents and carers need most • Improve access to rare disease information and psychosocial support • Reduce financial burden, including from out-of-pocket costs associated with care and treatment
15	Care and support for all Australians, including priority populations <ul style="list-style-type: none"> • Identify barriers to care and support for priority populations, including Aboriginal and Torres Strait Islander people, people from culturally and linguistically diverse backgrounds, those living in rural and regional Australia, and those with an undiagnosed rare disease or disability • Better support for all Australians to access specialised rare disease care and support, including genetic healthcare and psychosocial support

Rank	Theme
	<ul style="list-style-type: none"> • Improve access to specialised rare disease care and treatment for individuals and families living in rural and regional Australia, including by optimising the use of digital health services
16	Quality of life <ul style="list-style-type: none"> • Understand how rare disease impacts quality of life • Identify ways to improve quality of life for Australians living with rare diseases, their families and carers
17	Disability guidance and support <ul style="list-style-type: none"> • Assistance with the National Disability Insurance Scheme (NDIS) • Support with disability and aged care accommodation

*These themes were combined into one theme at the consensus workshops, *‘Development of and access to the best available treatments and cures, including clinical trials’*. All subthemes from both original themes were retained.

^These themes were combined into one theme at the consensus workshops, by *‘Support and information for reproductive choice’* being changed to a subtheme of *‘Genetic testing’*.