



**National Disability Insurance Scheme (NDIS)
Evidence Advisory Committee**

September 2025 Consultation

Rare Voices Australia Submission

November 2025

About Rare Voices Australia

[Rare Voices Australia](#) (RVA) is the national peak body for the estimated two million Australians living with a rare disease, nearly all of whom experience long-term impacts daily – impacts that meet the Australian Government’s definition of a disability. The disability impacts of living with a rare disease often are not recognised by policymakers.

RVA provides a strong, unified voice to advocate for policy as well as disability, health, and other systems that work for people living with a rare disease. RVA’s work is non-disease-specific and is based on the commonalities of approximately 7,000 different rare diseases. Our person-centred focus sees us working with all key stakeholders in the rare disease sector, including people living with a rare disease, governments, key peak bodies, researchers, clinicians, and industry. RVA advocates for the best outcomes for Australians living with a rare disease, and their families and carers.

RVA collaborates with over 100 rare disease groups/organisations in Australia that are consumer-led groups/organisations. A complete list of RVA Partner groups/organisations is available on [RVA’s website](#)

What Is a Rare Disease?

A disease is a condition with a specific pattern of clinical signs, symptoms, and findings, and is considered rare if it affects fewer than, or equal to, 5 in 10,000 people. There are approximately 7,000 different rare diseases and an estimated two million Australians live with a rare disease. Therefore, while the occurrence of individual rare diseases is uncommon, having a rare disease is relatively common.

Around 80% of rare diseases have a genetic origin and due to the hereditary nature of some rare diseases, multiple people within the same family can be impacted.

Rare diseases are often serious and progressive, exhibiting a high degree of symptom complexity, leading to significant disability, health, and psycho-social challenges.

Rare Disease Disability

Most people with a rare disease meet the Australian Government's definition of having a disability, which is defined as a “limitation, restriction or impairment, which has lasted, or is likely to last, for at least six months and restricts everyday activities.”^{1,2} **This includes the estimated 100,000 National Disability Insurance Scheme (NDIS) participants with severe and profound rare disease disability impacts.**

RVA has identified 5 categories of rare disease disability:

1. Neurological/neurodevelopmental disability
2. Progressive/degenerative disability
3. Fluctuating/episodic disability
4. Children with delayed development
5. Undiagnosed rare disease.

For key decision-makers at all levels, greater knowledge of rare diseases can facilitate more responsive and appropriate services for people living with a rare disease and their families and carers.

National Strategic Action Plan for Rare Diseases

RVA led the collaborative development of the Australian Government's [National Strategic Action Plan for Rare Diseases](#) (the Action Plan)³, the first nationally coordinated effort to address the needs of rare diseases in Australia. RVA is now leading the Action Plan's collaborative implementation on behalf of the rare disease sector. Aspects of the Action Plan specifically address the NDIS and the arbitrary and unhelpful line that is often drawn between health and disability. In particular, the Action Plan highlights the need for coordinated and integrated care (see Appendix 1).

The Action Plan is built on three foundational principles:

1. Person-centred care
2. Equity of access
3. Sustainable systems and workforce.

These principles directly support the recommendations in this submission.

Rare Voices Australia's Submission

Thank you for the opportunity to contribute to the foundational work of the newly established NDIS Evidence Advisory Committee (the Committee).

As the Committee begins its important task of reviewing evidence to provide advice to government on which supports should be included or excluded from NDIS funding, RVA writes to highlight critical considerations for engaging with the rare disease disability community.

People living with rare disease disability face unique challenges that must be acknowledged and addressed from the outset of the new Committee's processes, to ensure their equitable access to NDIS supports.

This submission outlines key issues and proposes practical solutions to improve the recognition of rare disease disability, promote more equitable and inclusive evidence-based approaches, and reduce the risk of necessary supports being excluded from NDIS funding. It also seeks to reduce the ongoing burden placed on individuals and caregivers, who are repeatedly required to explain the nuanced nature of their support needs in systems that do not yet fully reflect their lived experience.

Limitations of the Survey design

RVA is providing a written submission to this consultation rather than completing the Committee surveys as the surveys, which rely on selecting a type of disability and matching supports based on available evidence. This methodology assumes a linear and predictable relationship between diagnosis and support needs, an approach that does not reflect the episodic, fluctuating, and multi-dimensional nature of rare disease disability.

Individuals with rare disease disability may need different supports at different times for reasons that may not be directly linked to a diagnosis. A diagnosis-centric model risks excluding individuals with genuine but uncategorisable needs. A function-based, person-centred approach would better reflect these realities and ensure equitable access to supports.

Although the surveys aim to assess the safety and appropriateness of supports for certain types of disability, they fail to capture the layered and cumulative impacts of rare disease disability. These impacts often span multiple domains—physical, neurological, intellectual, cognitive, sensory, and psychosocial—intersecting in ways that can create significant barriers to support. For example, progressive physical decline may co-exist with emotional regulation challenges, sensory processing differences, and intellectual disability, fluctuating over time and compounding support needs.

Many rare diseases are progressive and require functional maintenance supports to prevent complications, preserve mobility, support respiratory and circulatory health, and maintain access to therapy. The survey design does not reflect these intersections, risking the exclusion of people whose needs cannot be neatly categorised while also overlooking the value of tailored, holistic approaches to complex rare disease disability.

Additionally, the surveys favour evidence drawn from large datasets focused on well researched and understood disability types. This approach consistently disadvantages rare disease cohorts and has

led to inequitable outcomes for people living with rare disease disability. The lack of traditional evidence should not be interpreted as a lack of need. Rare diseases are under-researched, and their diversity makes it nearly impossible to generate robust, diagnosis-linked evidence for every support type.

Recognition of Rare Disease Disability as a Distinct Cohort

People living with rare diseases often experience multiple, compounding disabilities that do not fit neatly into the existing NDIS disability types. These may include rare neurological impacts, fatigue-limiting symptoms, progressive muscular degeneration, skeletal and limb differences, sensory impairments, and complex psychosocial challenges, often co-existing and affecting multiple systems.

The disability categories listed by the Committee differ from those currently used by the broader [NDIS framework](#). For example, specific conditions such as amputation, arthritis, dementia, epilepsy, and language disorder are listed, while broader NDIS disability categories such as “Other Neurological”, “Other Physical” - categories often used to capture rare disease disability, are omitted.

The current Committee survey does not offer an option to identify rare disease disability, such as ‘Rare Disease/Condition/Syndrome’. Instead, individuals living with rare disease disability and their caregivers must select multiple categories from the available lists to try to convey the full impact of their condition. This process is often inaccurate or incomplete, and many are left resorting to the “Other” box. This masks the true nature and prevalence of rare disease disability and reinforces a sense of invisibility, making it harder to advocate for appropriate supports or policy changes.

RVA requests formal **recognition of rare disease disability as a distinct cohort**, and that a dedicated category for **“Rare Disease/Condition/Syndrome”** be included in all relevant assessment and data collection instruments, including Committee surveys and engagement activities.

RVA has consistently called for formal recognition of rare disease disability as a cohort and highlighted how the absence of reliable rare disease disability data is limiting effective government planning and undermines efforts to design equitable, responsive, and inclusive NDIS and disability supports. A dedicated rare disease disability category for the NDIS and this Committee would be a person-centred, trauma-informed step toward improving visibility, data accuracy, and policy outcomes.

Recommendation

1. The Committee should clarify why certain disability categories have been expanded while others remain general and explain how its categorisation framework aligns with the broader NDIS disability categories.
2. The Committee should formally recognise rare disease disability as a distinct cohort and include a dedicated category for “Rare Disease/Condition/Syndrome” in all relevant assessment and data collection instruments, including Committee surveys and engagement activities.

Impairment Categories

RVA questions the Committee's reliance on diagnosis categories without also incorporating impairment-based frameworks that align with the broader direction of the NDIS. The NDIS is designed to support individuals based on functional impairment, not solely just diagnosis. Impairment categories should also include a "multiple impairments" category to address the needs of individuals whose range of impairments precludes them from nominating a single primary impairment. This is particularly important for individuals with rare disease disability who may be undiagnosed or whose conditions do not fit neatly into existing diagnostic labels.

A diagnosis-only approach risks excluding or misrepresenting these individuals and undermines equitable access to supports. A function-based approach would better reflect the realities of rare disease disability, allowing for more flexible, responsive, and person-centred support planning.

Recommendation

3. That the Committee incorporate both diagnosis and impairment-based frameworks in its review to better reflect the diversity and complexity of disability in the rare disease community.

Inclusive Evidence Standards for Rare Disease Disability

The rare disease community is consistently disadvantaged by traditional research and evidence standards. Rare diseases affect small populations, making it difficult to conduct large-scale, statistically significant studies. Many conditions lack randomised controlled trials, or longitudinal data typically favoured in systematic reviews, resulting in systemic bias against rare and undiagnosed conditions.

Grey literature (e.g., reports, theses, guidelines) often excludes rare disease populations due to lack of funding, low awareness, or exclusion from broader disability studies. Systematic reviews often exclude real world evidence, case studies, and individualised clinical assessments, which are often the most relevant and useful forms of evidence for rare disease disability.

RVA advocates for the Committee to consider lived experience, clinical expertise, and contextual knowledge alongside published research and grey literature. Overlooking these sources risks overlooking evidence that identifies disability supports that are highly effective in practice, especially for conditions with variable or fluctuating symptoms.

There is also a significant evidence gap for individuals without a formal diagnosis. These people are often invisible in the research base leading to misunderstood, underassessed, and underfunded support needs. This perpetuates inequity for people living with rare disease disability. With limited peer-reviewed research and few professional bodies or conferences focused on rare disease disability, this community is often excluded from "evidence-based" funding decisions. As a result, effective supports may be deprioritised for individuals with rare disease disability simply due to absence of comparative data, not the absence of need.

RVA acknowledges that the Committee will be required to make **provisional recommendations** where the evidence base for a specific support is low quality, limited or still emerging.

In such cases, it is critical that the rare disease disability community understands how decisions will be made, who will be consulted, what types of evidence will be considered, and whether access to supports will continue if recommendations are deferred.

RVA is concerned that supports may be excluded from NDIS funding if further evidence is needed, but that evidence cannot be produced due to systemic barriers such as lack of funding, research infrastructure or interest. This risks deepening disadvantage for people with rare disease disability, who may miss out on essential NDIS supports.

Excluding supports due to evidence gaps risks perpetuating inequity and sidelining rare disease cohorts from policy reform, pricing reviews, and service design. This contradicts the principles of equity, person-centred care, and the Action Plan, which calls for inclusive, sustainable, and responsive systems.

To ensure recommendations reflect real-world needs, the Committee should engage directly and regularly with RVA as the national peak body for Australians living with a rare disease and our RVA Partners (rare disease groups/organisations) through the **Rare Disease Disability Network** (the RDDN). The RDDN is an RVA-led peer support and capacity building network for rare disease community-led groups/organisations and invited sector stakeholders. This collaboration offers access to lived experience, carer impacts, case studies, community consensus, and unpublished data and registries - essential to developing equitable and inclusive recommendations.

Recommendations:

The Committee should:

4. Explain how provisional recommendations will be made when evidence is limited, including what types of evidence will be considered and who will be consulted.
5. Publish all recommendations, including provisional ones, with a clear statement of the evidence considered, and any further evidence required.
6. Commit to inclusive evidence standards that recognise lived experience, professional judgement, and carer impact as legitimate and valuable forms of evidence, particularly for rare disease disability cohorts where lack of formal evidence is structural and systemic
7. Engage directly and regularly with RVA and RVA Partners, including through the Rare Disease Disability Network.

Conclusion

RVA urges the Committee to act on the recommendations outlined in this submission to improve visibility, equity, and access for people living with rare disease disability. By embedding these principles into its processes from the outset, the Committee can help build a more inclusive and sustainable NDIS—one that truly reflects the diversity and complexity of disability in Australia.

RVA welcomes all opportunities to collaborate and support the Committee in this important work, including through the Rare Disease Disability Network and our extensive partnerships across the rare disease sector.

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Appendix 1

Disability and the National Strategic Action Plan for Rare Diseases

Specific disability-related actions and implementation steps from the Action Plan include:

Action 2.1.1: Provide rare disease care and support that is integrated, incorporating clear pathways throughout health, disability, and other systems.

Implementation

2.1.1.2. To reduce fragmented care, ensure policy meets people's full range of needs, including health, disability and education. Support this work with a cross-jurisdictional, cross-sectoral working party.

Action 2.1.2: Build a broad range of care and support services that are responsive to the changing needs of people living with a rare disease and their families.

Implementation

2.1.2.1. Develop an accessible multi-purpose digital repository, incorporating elements targeted at the workforce that supports people living with a rare disease. With access to adequate information, health care and social support professionals will be equipped to support people living with rare disease and their families to navigate health, disability, and other systems.

2.1.2.3. Through regular stakeholder consultations, determine strategies to improve access to rare disease care and support services for Aboriginal and Torres Strait Islander people, those with CALD backgrounds, those living in rural and remote areas, and other priority populations.

References

1. Australian Government. Australian Public Service Commission. Definition of disability. September 2019. Available from: <https://www.apsc.gov.au/working-aps/diversity-and-inclusion/disability/definition-disability> [Accessed November 2025]
2. Australian Bureau of Statistics. Disability, ageing and carers, Australia: Summary of findings. July 2024. Available from: <https://www.abs.gov.au/statistics/health/disability/disability-ageing-and-carers-australia-summary-findings/latest-release> [Accessed November 2025]
3. Commonwealth of Australia. Department of Health. *National Strategic Action Plan for Rare Diseases*. Canberra; 2020. Available from: <https://www.health.gov.au/sites/default/files/documents/2020/03/national-strategic-action-plan-for-rare-diseases.pdf> [Accessed November 2025]