Senate Standing Committees on Community Affairs
Community Affairs References Committee Inquiry
Into Epilepsy in Australia

May 2026

Rare Voices Australia Submission

About Rare Voices Australia

Rare Voices Australia (RVA) is the national peak body for Australians 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 means we work with key stakeholders in the rare disease sector, including people living with a rare disease, governments, key peak bodies, researchers, clinicians, and industry. We collaborate with over 100 consumer-led rare disease groups/organisations (RVA Partners) across Australia to provide a strong, unified voice.

RVA advocates for the best outcomes for Australians living with a rare disease, which includes families and carers.

What Is a Rare Disease?

A disease is considered rare if it affects fewer than, or equal to, 5 in 10,000 people [1]. There are approximately 7,000 different rare diseases and an estimated two million Australians live with a rare disease. While individual diseases may be rare, the total number of Australians living with a rare disease is not.

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[1].

Rare diseases are:

- Recognised globally by the United Nations and World Health Organization (WHO) as a critical health and equity priority. The WHO is currently developing a 10-year Global Action Plan to address the common challenges experienced by people living with a rare disease. [2]
- Recognised by the World Economic Forum as driver of healthcare sustainability and innovation.[3]

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)[1], the first nationally coordinated effort to address rare diseases in Australia. RVA is now leading the Action Plan's collaborative implementation on behalf of the rare disease sector.

The Action Plan is built on three foundational principles:

- Person-centred
- Equity of access
- Sustainable systems and workforce.

These principles directly support the recommendations in this submission.

Rare Disease and Epilepsy

The relationship between epilepsy and rare disease is significant. Evidence indicates that approximately 44% of people diagnosed with epilepsy have an underlying rare genetic condition [4]. The Epilepsy Foundation Australia recognises this substantial overlap and provides targeted information on rare and genetic epilepsies [5].

Rare diseases are frequently serious, complex, and progressive in nature. They are characterised by high levels of clinical heterogeneity and symptom burden, resulting in enduring health and psychosocial impacts[1]. These challenges are experienced in rare diseases associated with epilepsy. For example, genetic epilepsies and developmental epileptic encephalopathy, where individuals frequently experience intellectual disability, and multiple co-morbidities, resulting in highly complex care needs. Epilepsy associated with rare genetic diseases reflects the broader systemic challenges faced across the rare disease landscape, including:

- Limited awareness and clinical knowledge of rare genetic conditions that have associated epilepsies. This results in systems failing to account for ongoing instability, regression risk and deterioration frequently associated with these conditions.
- Delayed, missed, or incorrect diagnosis.
- Restricted access to specialist expertise and multidisciplinary care.
- Limited availability of effective treatments.
- Fragmented and poorly coordinated care across health, disability, and social systems. This includes transition to adult services that frequently lack knowledge and experience in managing complex epilepsy alongside complex disability.
- Inadequate access to psychosocial support for individuals and families.
- Lack of comprehensive data collection and national research coordination.

These systemic gaps result in poorer health outcomes, increased burden on families and caregivers, and avoidable long-term costs to the health system.

Specific Needs of People with Rare Genetic Epilepsy

People living with epilepsy associated with an underlying rare genetic condition have distinct and highly complex needs compared to those with more common forms of epilepsy.

They require:

- Equitable and timely access to genomic testing, including Whole Genome Sequencing (WGS), to enable early and accurate diagnosis.
- Access to specialist expertise and coordinated, multidisciplinary care lifespan models that are inclusive and address the additional complexity involved with rare disease related epilepsies.
- Integrated care across systems, including health, disability, education, and employment.
- Embedded mental health and psychosocial support for individuals and families.

- Access to emerging and precision therapies, including participation in clinical trials.
- A national research ecosystem capable of supporting ultra-rare populations and translational innovation.

Without this support, individuals face prolonged diagnostic journeys, inappropriate treatments, and significant inequities in care.

Response to Terms of Reference

RVA acknowledges the Inquiry's Terms of Reference and notes that the Action Plan already provides a comprehensive framework that addresses the systemic barriers experienced by people living with a rare disease.

This submission therefore focuses on targeted actions to implement the Action Plan in ways that directly address epilepsy associated with rare genetic conditions.

RVA emphasises that this cohort should be recognised as a distinct group within the epilepsy population, due to the additional complexity, severity, and service requirements associated with rare genetic disease.

Recommendation 1: Invest in Nationally Networked Rare Disease Centres of Expertise

RVA calls for Commonwealth investment into the development of nationally networked Rare Disease Centres of Expertise (CoE). The Action Plan identified networks of CoE as a key implementation mechanism[1]. It also highlights international exemplars of best practice, including the European Reference Networks as a model that could address the challenges of providing expert information care to a small geographically dispersed population, such as those with epilepsy related to rare diseases.[1]

CoEs area an optimal rare disease model of care with international exemplars outlined in the Action plan. In the context of rare and genetic epilepsies they would:

- Develop workforce capacity to respond to the complex challenges of rare disease related epilepsy, including rare genetic epilepsies and developmental epileptic encephalopathy (Action Plan Priority 1.3: Develop a national rare disease workforce strategy that responds to current and future demands, including the impact of genomics)[1].

- Facilitate equitable and early diagnosis of rare diseases associated with epilepsy (Action Plan Action 2.2.3: People with an undiagnosed rare disease are identified and have priority access to the most appropriate specialised and expert diagnostic response)[1].

- Provide an expert reference resource to facilitate primary care providers delivering expert informed care, which will reduce both cost and travel burden as well as

challenges in accessing specialist care and ensuring family-centred care. (Action Plan Priority 2.1: Provide rare disease care and support that is integrated and appropriate for all Australians living with a rare disease, while being both person and family-centred.)[1]

- Lead the development of cross sector integrated care models similar to the Clinical Centre of Expertise for Rare and Undiagnosed Diseases, ensuring that care integrates all systems that families will need to navigate living with epilepsy associated with rare conditions, reducing the burden on families. (Action Plan Action 2.1.1: Provide rare disease care and support that is integrated, incorporating clear pathways throughout health, disability and other systems)[1].

- Provide mental health and wellbeing support, informed by rare disease knowledge, for people living with epilepsy associated with a rare disease and their carers and family . (Action Plan Priority 2.5: Integrate mental health, and social and emotional wellbeing, into rare disease care and support)[1].

- Facilitate innovative translational research, ensuring Australians with epilepsy associated with rare disease have early access to emerging treatments integrated with ongoing clinical care and support. (Action Plan Priority 3.4: Translate research and innovation into clinical care; clinical care informs research and innovation, and Action 3.2.4: Building on existing initiatives, continue to foster an environment conducive to clinical trials for rare diseases taking place in Australia)[1].

Recommendation 2: Health Technology Assessment Reform

RVA strongly supports the full implementation of the recommendations from the Health Technology Assessment (HTA) Review, with a focus on:

- Incentivising innovation for rare and ultra-rare conditions.
- Addressing high unmet clinical need.
- Reducing delays in access to life-changing therapies.

Reform is essential to ensuring Australians living with rare genetic epilepsies have timely and equitable access to emerging treatments, including gene and precision therapies in alignment with Action Plan Priority 2.4: Enable all Australians to have equitable access to the best available health technology[1].

Recommendation 3: Investment in Rare Disease Research and Data Collection

RVA recommends that the Medical Research Future Fund (MRFF) develops a rare disease funding stream to address key gaps in rare disease research and data collection. This aligns with Action 3.2.1 of the Action Plan: Develop a national research strategy for rare diseases, to keep pace with genomic advancements, precision medicine and innovation, and Priority 3.1: Enable coordinated and collaborative data collection to facilitate the monitoring and cumulative knowledge of rare diseases, informing care management, research and health system planning. [1]

Investment in rare disease research:

- Addresses one of the most significant areas of unmet clinical need.
- Drives innovation in genomics, precision medicine, and advanced therapeutics.
- Generates system-wide benefits extending beyond rare disease populations.

Conclusion

The Australian Government's Action Plan provides a clear and comprehensive policy framework to address the needs of Australians living with epilepsies associated with rare genetic diseases. However, Action Plan implementation remains incomplete.

Targeted Commonwealth investment, particularly in nationally networked Rare Disease CoE, HTA reform, and MRFF funding:

- Reduces diagnostic delays.
- Improves care coordination.
- Enables equitable access to emerging therapies.
- Delivers better health and psychosocial outcomes.

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