SCN2A AUSTRALIA

Senate Inquiry Submission

Developmental and Epileptic Encephalopathies

Evidence, priorities, and recommendations

Submitted to	Senate Standing Committee on Community Affairs
Inquiry	Inquiry into Epilepsy in Australia
Submitting organisation	SCN2A Australia
Date	May 2026
Contact	Kris Pierce, Founder and Director
Email	info@scn2aaustralia.org.au

ABN 70 665 788 161 · ACN 665 788 161

scn2aaustralia.org

Executive summary

Rare and complex epilepsies, including developmental and epileptic encephalopathies (DEEs) and genetic conditions such as SCN2A-related disorders, are defined by drug-resistant seizures, multimorbidity, premature mortality, and lifelong care needs. They are also the segment of the epilepsy population where the therapeutic pipeline is most active, and where early, accurate diagnosis determines whether treatment helps or causes harm.

Three independent processes within twelve months reached the same conclusion: current Australian systems are not meeting the needs of this population. The DEE Roundtable 2026(1), the IBE Rare Epilepsy Leaders Global Convening, and the published Global Epilepsy Needs Study (GENS I) each identified the same structural failures. Families absorb the coordination function across health, disability, and education systems that do not communicate with each other. Diagnostic delay produces clinical harm. Adult care pathways do not yet exist for the first generation of genetically diagnosed DEE patients now reaching adulthood. Caregiver mental health, sibling impact, and family financial wellbeing are not recognised as policy priorities, despite documented evidence.

The economic case mirrors the clinical one. Approximately 300 children are newly diagnosed with a DEE in Australia each year, an incidence of around 1 per 1,000 [2–4]. These children have exceptionally high healthcare needs. In a recent Australian sample, 61 per cent were hospitalised within a 12-month period, with an average admission of 23.2 days [5]. At an inflation-adjusted cost of $3,848 per paediatric bed-day, annual hospitalisation costs for this incident cohort sit at roughly $26.8 million [7,8]. Indirect costs to families, including lost productivity, transport, and accommodation, conservatively add a further $2.12 million per year at a minimum of $500 per day [9].

Over time, the burden compounds across health, disability, education, and social systems. Without earlier diagnosis, coordinated care, and access to appropriate treatment and supports, families continue to carry significant lifelong impacts that extend well beyond the individual living with epilepsy. The cumulative effect is substantial, affecting not only health outcomes, but caregiver workforce participation, family wellbeing, and broader system sustainability.

About SCN2A Australia

SCN2A Australia is a national charity representing people living with SCN2A-related disorders and their families. SCN2A is one of the leading genetic causes of severe childhood epilepsy, autism, and developmental and intellectual disability. The organisation provides peer support, partners on and funds research, contributes to national and international policy, and advocates for systemic reform on behalf of the Australian DEE community.

Our authority to contribute to this inquiry rests on three things: lived expertise, recent national and international engagement, and evidence.

Lived expertise sits at the centre of the organisation. Our board is led by professionals who also have lived experience as caregivers of a child with SCN2A, and who provide broader strategic input through senior national advisory roles, including Deputy Chair of the HTA Consumer Consultative Committee, member of the MSAC Evaluation Sub-Committee, and member of the Genomics Australia Advisory Council.

Recent organisational engagement includes convening the Rare Epilepsy Leaders Global Convening in London with the International Bureau for Epilepsy (April 2026), and delivery of the National DEE Roundtable 2026. These forums place SCN2A Australia at the centre of current Australian and international policy dialogue on rare epilepsies.

The evidence base for this submission draws on the SCN2A Australia families survey (April 2026) [6], the outcomes of the DEE Roundtable 2026 [2], peer-reviewed research, and the international dialogue emerging through the IBE. The survey sample reflects the ultra-rare nature of SCN2A-related disorders; testimony is used as qualitative evidence of recurring patterns within our community.

Central needs

- Formally recognise rare and complex epilepsies as a distinct priority population.
- Fund a national DEE registry covering diagnosis, multimorbidity, treatment response, and mortality.
- Ensure early, comprehensive, reimbursed genomic testing at first presentation.
- Fund whole-of-life, multi-sector models of care across health, disability, and education.
- Reform HTA frameworks to accommodate variant-level heterogeneity and precision therapies.

1. SCN2A as a representative DEE: clinical and policy profile

SCN2A encodes the Nav1.2 sodium channel subunit, essential for normal neuronal firing. Pathogenic variants produce a clinical spectrum ranging from neonatal-onset intractable epilepsy with profound developmental impact, to autism and intellectual disability with little or no seizure activity. Clinical heterogeneity is mechanism-dependent: gain-of-function variants typically cause early-onset, drug-resistant seizures; loss-of-function variants more often produce autism and intellectual disability. The distinction is clinically critical, because sodium channel blockers, a standard antiseizure medication class, can worsen seizures in patients with loss-of-function variants. Molecular diagnosis is not optional; it determines whether treatment is safe [9,10].

The consequences of delayed or missed diagnosis are not abstract. One family in the SCN2A Australia families survey reported:

> *We pushed for seven years to get genetic testing. We got rejected from the genetic clinic so many times. Our son only had an autism diagnosis but the therapies for autism were unsuitable for him. He only got diagnosed at 9 years old. Our son was using the wrong medication for SCN2A. We got so much judgment from his behaviours as an 'autistic' child; our family's situation would have been so much easier if we had known from a young age his true diagnosis.*
>
> — SCN2A caregiver, SCN2A Australia families survey, 2026

This is direct evidence of treatment harm from missed diagnosis. It is not a single case; survey responses describe diagnostic delays of 7 to 14 years across the cohort.

SCN2A-related disorders rarely present as epilepsy alone. Most affected children have drug-resistant seizures combined with intellectual disability, autism, movement disorders, hypotonia, feeding difficulties, sleep disruption, cortical visual impairment, respiratory vulnerability, and autonomic dysfunction. Published cohort data report mortality of 13 to 15

per cent in SCN2A-related DEE, comparable to Dravet syndrome, with causes including SUDEP, status epilepticus, respiratory illness, severe infection, and autonomic dysfunction. Population-level mortality across the full SCN2A spectrum has not been systematically quantified in Australia. The surveillance gap is itself a policy problem: without data, the system cannot assess whether interventions are working (10,11,12).

SCN2A illustrates structural problems common to the wider DEE population: mechanism-dependent treatment requiring molecular diagnosis, multimorbidity requiring coordinated care, a precision therapeutic pipeline requiring adaptive regulatory and HTA frameworks, and surveillance gaps that impede both clinical management and research. The policy task applies across 900+ DEE genes.

2. Convergent evidence: national consensus and international evidence

The DEE Roundtable 2026, convened by SCN2A Australia in May 2026, brought together families and lived experience advocates, neurologists, epileptologists, clinical geneticists, researchers, allied health professionals, policymakers, HTA specialists, patient advocacy organisations, and pharmaceutical and biotechnology industry representatives. Three priorities achieved strong support:

- Formal recognition of rare and complex epilepsies, including DEEs, as a distinct priority group, not a subset of general epilepsy.
- A national DEE registry with standardised data elements covering genetic diagnosis, multimorbidity, treatment response, and mortality.
- Coordinated models of care across health, disability, and education, with a single care coordination function per family across the whole of life.

Participants also called for a dedicated DEE workforce, including nurse navigators, and a national DEE action plan modelled on cancer-style standards of care. A full report will be published and supplied to the Committee.

The April 2026 IBE Rare Epilepsy Leaders Global Convening, in which SCN2A Australia participated, identified shared international priorities: a common advocacy framework for rare and complex epilepsies, caregiver mental health, access to non-pharmacological care, consistent seizure emergency protocols, and patient-reported outcome measures that capture outcomes meaningful to families beyond seizure frequency.

The convening also advanced planning for the Global Epilepsy Needs Study II (GENS II), which focuses on rare and complex epilepsies. GENS I, published in Epilepsia Open in 2026 [13], surveyed 5,296 people across 15 countries, including Australia, and found that caregivers of people with complex epilepsy were more than twice as likely as caregivers of people with non-complex epilepsy to require respite and inclusive social supports. The study identified significant gaps in tailored services, multidisciplinary care, respite, and transition planning.

Australian peer-reviewed evidence reinforces these findings. Kelada et al. found that 69 per cent of DEE families presented to emergency departments within 12 months, most more than once, and that caregivers prioritised improved hospital care and psychological support; the authors concluded that DEE models of care must include care coordination, knowledgeable clinicians, and psychological support [5]. Pierce et al. documented the cumulative toll on caregiver wellbeing in Australian carers of children with rare epilepsies, including chronic

isolation, erosion of self-efficacy, and loss of social connection alongside the everyday demands of complex care [13].

Three independent processes have reached convergent priorities for DEEs within twelve months. The remaining task is implementation, in line with the WHO Intersectoral Global Action Plan on Epilepsy and Other Neurological Disorders (IGAP) [14], the National Strategic Action Plan for Rare Diseases [15], and the National Strategic Framework for Chronic Conditions [16], all of which Australia has endorsed but which lack a DEE-specific implementation plan.

3. Recommendations

Recommendations are organised into five focus areas drawn from the Committee's Terms of Reference. They are cumulative and interdependent: registry data underpins workforce planning, HTA reform depends on diagnostic accuracy, and family support is part of the model of care, not separate from it.

Focus area 1: Diagnosis and genomic testing access

Terms of Reference 1, 2

For rare and complex epilepsies, the diagnostic pathway determines treatment safety. Without molecular diagnosis, clinicians cannot reliably distinguish gain-of-function from loss-of-function variants, and standard antiseizure medications can cause harm. The SCN2A Australia families survey reports diagnostic delays of 7 to 14 years, repeated rejections by clinical genetics services, and treatment with medication contraindicated for the molecular subtype [6]. Geographic, workforce, and cultural barriers compound at every step. One rural family described 9 hours of driving each way for neurology appointments and a 6-month wait for a sedated EEG.

> **R1. Ensure access to early, comprehensive, reimbursed genomic testing at first presentation for any child with suspected DEE, early-onset epilepsy of unknown cause, or epilepsy associated with intellectual disability or autism, before multiple failed antiseizure medication trials.**

- National MBS item review for paediatric epilepsy genomic testing.
- Build state-level clinical genetics service capacity to meet demand at first presentation.
- Standardised national referral pathways from paediatricians and GPs.
- Variant classification and curation infrastructure, with sustained Australian participation in international genomic databases.

> **R2. Reduce geographic and cultural inequity in diagnostic access through funded telehealth pathways and co-designed culturally safe resources, particularly for regional, remote, Aboriginal and Torres Strait Islander, and culturally and linguistically diverse families.**

- Funded telehealth links between regional families, paediatricians, GPs, and tertiary epilepsy and clinical genetics services.
- Decision-support and diagnostic resources co-designed with Aboriginal and Torres Strait Islander and CALD lived experience advocates.

R3. Commission a national workforce strategy for paediatric neurology and clinical genetics, including modelling of future DEE demand, training pipeline requirements, and equitable rural workforce distribution.

- Informed by the Rare Metabolic Disease Workforce White Paper and DEE-specific demand modelling.
- Funded nurse navigator and care coordinator roles.

Focus area 2: Coordinated multi-sector care across health, disability, and education

Terms of Reference 2, 4

System fragmentation is most damaging at the interface of health, disability, education, and adult services. Families become the default coordinators of care across systems with different eligibility, funding, and accountability mechanisms. NDIS access is inconsistent: cognitive, behavioural, psychosocial, and complex medical needs are variably recognised; seizure management equipment and respite are disputed; and administrative failures impose direct financial cost on families with no mechanism for accountability. Education is a particular gap. As one family in the SCN2A Australia families survey described it:

> *Schooling. Keeping children safe and happy is not enough. They also need daily therapy and appropriate education.*
>
> — SCN2A Australia families survey, 2026

Adult care pathways are not yet established. The first generation of genetically diagnosed DEE patients is now entering adulthood without equivalent adult systems. The most senior person living with SCN2A-related disorder in our community is 24 years old. The survey response describing his care is the clearest evidence in the dataset of the adult care gap:

> *Our son has SCN2A-related disorder, one of the most severe developmental and epileptic encephalopathies. He requires 24-hour care, seven days a week. He cannot be left safely in the community without support. There is no coordinated system supporting us; there is only my husband and me. Life is managed, not lived.*
>
> — SCN2A Australia families survey, 2026

R4. Fund whole-of-life, multidisciplinary, family-inclusive DEE models of care coordinated across health, disability, education, psychosocial, and palliative care, with a single accountable care coordination function per family.

- Funded nurse navigator or care coordinator per family across the whole of life.
- Integrated case conferencing mechanism linking treating neurologist, allied health, school, NDIS, and primary care.
- Embedded patient-reported and caregiver-reported outcome measures.
- Standardised individual seizure emergency care protocols across emergency departments, primary care, schools, and community settings.

R5. Establish a specialised NDIS pathway for developmental and epileptic encephalopathies (DEEs) and other rare epilepsies, recognising that the current framework is structurally misaligned with conditions characterised by

developmental regression, treatment resistance, lifelong complexity, and evolving care needs.

This pathway should include:

- Dedicated direct planners with specialist training in DEEs and rare genetic epilepsies, consistent with specialist planner models already established for other progressive neurological conditions.
- Mandatory proactive reassessment and flexible funding mechanisms that respond to deterioration, hospitalisation, or significant functional change, rather than relying on static plans and automatic roll-overs.
- Formal recognition that severe genetic epilepsies constitute permanent disability at diagnosis and should not require repeated proof of treatment exhaustion or functional decline to access support once diagnosed.
- Funding pathways for skilled in-home clinical supports, including seizure management, airway support, overnight monitoring, and registered nursing where clinically required, recognising that the current 'medical versus disability' distinction places families and participants at risk.
- An agreed mechanism for ongoing consultation, feedback, and co-design with the DEE community through established paediatric neurology, epilepsy, palliative care, and lived experience working groups, ensuring NDIS policy, operational guidance, and planning frameworks remain responsive to the evolving clinical and psychosocial realities of developmental and epileptic encephalopathies.
- Structured transition protocols for movement between paediatric and adult systems, recognising the substantial care discontinuity experienced by adults living with childhood-onset epileptic encephalopathies and the limited expertise within adult disability and neurology systems for these conditions.

R6. Establish nationally coordinated adult care pathways for people living with developmental and epileptic encephalopathies (DEEs), including structured transition from paediatric to adult neurology, disability, psychosocial, palliative, and community care services.

- Adult DEE clinical leads identified within each jurisdiction to support continuity of care, multidisciplinary coordination, and expertise in rare and complex epilepsies.
- Transition protocols commencing no later than age 14, with formal joint planning processes between paediatric and adult services to prevent care fragmentation and loss of support.
- Recognition that many current adult patients with DEEs have aged out of paediatric systems without equivalent adult systems being established; immediate remedial investment and service provision is required for this cohort.
- Development of a DEE-specific decision-support and care pathway framework, endorsed in partnership with Palliative Care Australia, recognising the progressive, life-limiting, and high-burden nature of many DEEs.
- Integration of psychosocial, palliative, and bereavement support pathways following significant deterioration, recurrent status epilepticus, intensive care admissions, and sudden unexpected death in epilepsy (SUDEP), recognising the cumulative trauma experienced by families living with lifelong severe epilepsies.

Focus area 3: Family and caregiver support as a recognised policy priority

Terms of Reference 2, 4

Caregiver wellbeing is not currently recognised as a core policy priority despite documented evidence of severe, cumulative, and predictable harm. GENS I found caregivers of people with complex epilepsy were more than twice as likely to require respite and inclusive social supports than caregivers of people with non-complex epilepsy. Kelada et al. (2025) identified parent psychological support as a top family-identified priority for service improvement. The SCN2A Australia families survey describes withdrawal from paid employment over decades, no superannuation accrual, deferred home ownership, chronic poor mental health, anticipatory grief, repeated exposure to medical trauma, and structural exclusion from carer payments due to means and asset testing [6].

Families living with DEEs frequently navigate life-limiting uncertainty, recurrent status epilepticus, intensive care admissions, fear of SUDEP, progressive decline, and medically complex decision-making over many years. Despite this, access to psychosocial and palliative care supports remains inconsistent, late-stage, or absent altogether. Palliative care continues to be poorly understood within DEEs, despite its recognised role in supporting quality of life, symptom burden, caregiver wellbeing, advance care planning, and bereavement support across the trajectory of serious neurological conditions.

Sibling impact is consistently raised but rarely addressed in policy. Two families in the survey described it directly:

> *Extreme violent behaviours; son attacks sibling and sibling now treated for trauma.*

> *Constant vigilance, constant demands and problems to navigate, frequent damage and breakages, sleeplessness, constant costs, constant exhaustion, relative neglect of other children in the family.*
>
> — SCN2A Australia families survey, 2026

Caregiver mental health, sibling impact, anticipatory grief, bereavement support, and family financial security are not adjuncts to DEE care. They are predictable consequences of the current system and require formal recognition within health, disability, and palliative care policy frameworks.

> **R7. Recognise caregiver mental health and psychosocial wellbeing as a formal policy priority within DEE models of care, with integrated psychological, peer, and palliative care support embedded across the lifespan, including explicit inclusion of siblings and bereaved families.**

- Funded caregiver mental health screening at key clinical care points, including diagnosis, major deterioration, transition periods, ICU admissions, and post-bereavement.
- Early integration of paediatric and adult palliative care approaches within DEE care pathways, recognising the cumulative burden, uncertainty, and life-limiting nature of many DEEs.

- Sibling-specific psychosocial support, recognising trauma, chronic stress, behavioural exposure, and neglect documented in the SCN2A Australia families survey and broader DEE evidence.
- Peer support funded as a core component of care, not an adjunct service.
- Access to structured bereavement support following SUDEP, status epilepticus-related death, treatment complications, or progressive decline.

> **R8. Address the structural financial harm imposed on DEE families by current carer payment, NDIS, respite, and employment arrangements.**

- Review means and asset testing of carer payments for households where one parent has withdrawn from paid employment to provide 24-hour care.
- Recognise lost superannuation accrual, reduced workforce participation, and deferred home ownership as measurable policy consequences requiring remediation.
- Commission a national economic analysis of drug-resistant epilepsy and DEEs, including direct health costs, carer burden, lost productivity, out-of-pocket costs, informal care, and long-term financial disadvantage.

Focus area 4: Research infrastructure and clinical trial access

Term of Reference 5

Commonwealth research investment in epilepsy does not reflect the burden, complexity, or therapeutic opportunity of rare and complex epilepsies. The DEE therapeutic pipeline is advancing rapidly through antisense oligonucleotides, gene therapies, precision pharmacology, and emerging N-of-1 approaches. Australia has internationally recognised expertise in genomics, HTA, clinical research, and lived experience engagement that could support leadership in this field; yet investigator-led DEE research is constrained by small populations, fragmented datasets, limited registry infrastructure, and the challenge of demonstrating treatment effect across heterogeneous natural histories.

Australian SCN2A experience also illustrates a structural issue in translational research governance. Australian families contributed biological samples, longitudinal natural-history data, and years of research participation, which supported mechanistic understanding and therapeutic development. These community-generated assets contributed to development of both small-molecule and antisense oligonucleotide candidates. No Australian clinical trial sites were established across the development pathway, and data generated through community participation has not been consistently shared back with participating families or the broader research community. As precision therapies for rare epilepsies expand, the same pattern is likely to recur unless Australia establishes clearer expectations regarding stewardship of community-generated research assets, equitable trial participation, and national access pathways. Adding to this, the US-based company has recently advised our Board that 'there are no immediate global registration plans'.

> **R9. Establish a national DEE registry and supporting research infrastructure, including longitudinal natural-history data, biobanking, variant curation, interoperable datasets, and platform-trial capability suitable for ultra-rare populations.**

- Governance enabling research linkage and ongoing surveillance.

- Standardised data elements covering genetic diagnosis, multimorbidity, treatment response, healthcare utilisation, NDIS supports, and mortality.
- Held in partnership with patient organisations, with lived experience embedded at every level of governance.

R10. Establish a dedicated Commonwealth research funding stream for rare and complex epilepsies, spanning basic science, natural history, biomarkers, clinical trials, implementation science, and health systems research, with mandated lived experience leadership.

R11. Establish a national governance framework for community-generated research assets in rare disease and precision medicine, and adapt HTA frameworks for variant-level heterogeneity and precision therapies.

- Timely de-identified data sharing back to participating families and the research community.
- Transparent communication and expectations regarding Australian regulatory and access pathways, where Australian patients and infrastructure contributed materially to therapeutic development.
- HTA guidance on natural history data, surrogate endpoints, and real-world evidence in lieu of randomised controlled trial data for variant-defined populations.
- Streamlined pathways into compassionate access, Named Patient Programmes, and clinical trials, including expedited regulatory pathways for serious genetic conditions with no approved therapies.

Focus area 5: Recognition, action plan, and lived experience leadership

Term of Reference 6

Recognition is the precondition for everything else in this submission. Without it, DEEs remain a subset of general epilepsy in policy terms, ineligible for the dedicated workforce, registry, action plan, and HTA reform that the evidence base, the DEE Roundtable consensus, and international convergence all support.

R12. Formally recognise rare and complex epilepsies, including DEEs, as a distinct priority population within Commonwealth health, disability, education, and research policy, separate from epilepsy as a general category.

R13. Develop a national DEE action plan with measurable targets, modelled on coordinated chronic disease and cancer frameworks.

- Targets covering diagnosis, multidisciplinary care access, transition and adult care, NDIS adequacy, research investment, clinical trial participation, and mortality.
- Public reporting against targets.
- Domestic implementation aligned with WHO IGAP through the Epilepsy and other Neurological Disorders Alliance (ENDA).

R14. Establish a standing mechanism for lived experience leadership within DEE policy, research, and implementation structures arising from this inquiry, with defined governance roles and decision-making authority beyond consultative participation.

Closing

The evidence base is established. The policy frameworks already exist. The precision medicine pipeline is advancing. What remains is a Commonwealth decision to implement the structural reforms required to align Australian systems with current science, international direction, and the documented experience of families living with rare and complex epilepsies.

We would welcome the opportunity to appear before the Committee. SCN2A Australia will provide the DEE Roundtable 2026 report and supporting materials to the Committee on publication.

References

1. Pierce K, Howell KB, Milne K, Palmer E, Deacon N, Cross M, Duffy L. DEE Roundtable 2026: from insights to impact discussion paper. SCN2A Australia, Rare Insight Advisory, and DEE Roundtable Steering Committee; 2026.
2. Scheffer IE, Bennett CA, Gill D, de Silva MG, Boggs K, Marum J, et al. Exome sequencing for patients with developmental and epileptic encephalopathies in clinical practice. Developmental Medicine and Child Neurology. 2023;65(1):50–57.
3. Palmer EE, Schofield D, Shrestha R, Kandula T, Macintosh R, Lawson JA, et al. Integrating exome sequencing into a diagnostic pathway for epileptic encephalopathy: evidence of clinical utility and cost effectiveness. Molecular Genetics and Genomic Medicine. 2018;6(2):186–199. doi:10.1002/mgg3.355
4. Howell KB, Eggers S, Dalziel K, Riseley J, Mandelstam S, Myers CT, et al. A population-based cost-effectiveness study of early genetic testing in severe epilepsies of infancy. Epilepsia. 2018;59(6):1177–1187. doi:10.1111/epi.14087
5. Kelada L, Best S, Pierce K, Allen M, Cobb J, Berens K, Goranitis I, Palmer EE, Scheffer IE, Howell KB. Fulfilling the needs of caregivers in delivering health services to children with developmental and epileptic encephalopathies. European Journal of Paediatric Neurology. 2025;54:147–158. doi:10.1016/j.ejpn.2025.01.007
6. SCN2A Australia. Developmental and epileptic encephalopathies: Australian family experiences survey 2026 [unpublished online survey]. SCN2A Australia; 2026.
7. Independent Hospital Pricing Authority. National Hospital Cost Data Collection Cost Report: Round 20, Financial Year 2015–16. Sydney: IHPA; 2018.
8. Australian Bureau of Statistics. Consumer Price Index, Australia. Canberra: ABS; 2024. Available from: https://www.abs.gov.au/statistics/economy/price-indexes-and-inflation/consumer-price-index-australia/latest-release
9. Demetriou EA, Boulton KA, Thapa R, Sun C, Gilroy J, Bowden MR, et al. Burden of paediatric hospitalisations to the health care system, child and family: a systematic review of Australian studies (1990–2022). The Lancet Regional Health: Western Pacific. 2023;40:100878. doi:10.1016/j.lanwpc.2023.100878
10. Wolff M, Johannesen KM, Hedrich UBS, et al. Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. Brain. 2017;140(5):1316–1336.
11. Howell KB, McMahon JM, Carvill GL, et al. SCN2A encephalopathy: a major cause of epilepsy of infancy with migrating focal seizures. Neurology. 2015;85(11):958–966. doi:10.1212/WNL.0000000000001926
12. Sanders SJ, Campbell AJ, Cottrell JR, et al. Progress in understanding and treating SCN2A-mediated disorders. Trends in Neurosciences. 2018;41(7):442–456.
13. Baker GA, Bagga SK, Walsh D, Nolan C, Sharma S, Hooker C, Gonzalez Parrao P, Sofia F, Cross JH, Bertone MM, García I, Janneh AJ, Kukla A, Montero EV, Ding D, Friedrich L, Pinto LF, Singh G, Triki CC, Raj L, Reese A; GENS Collaborators. The Global Epilepsy Needs Study (GENS): a mixed-methods, multi-country exploration of the unmet psychosocial and everyday needs of people with epilepsy. Epilepsia Open. 2026 Apr 15. doi:10.1002/epi4.70237.

14. Pierce K, Murphy JB, Robertson EG, Khan JR, Bullock S, O'Loughlin CB, Loden M, McIntosh R, Beavis E, Roberts N, Palmer EE, Lingam R. 'Just realising that I wasn't alone… was profound': a mixed-methods evaluation of a pilot peer-to-peer wellbeing program for carers of children with rare epilepsies. Orphanet Journal of Rare Diseases. 2025;20(1):524. doi:10.1186/s13023-025-04036-0.

15. World Health Organization. Intersectoral Global Action Plan on Epilepsy and Other Neurological Disorders 2022–2031. Geneva: WHO; 2022.

16. Australian Government Department of Health. National Strategic Action Plan for Rare Diseases. Canberra: Commonwealth of Australia; 2020.

17. Australian Government Department of Health. National Strategic Framework for Chronic Conditions. Canberra: Commonwealth of Australia; 2017.