InGeNA submission to the Senate Inquiry into Epilepsy in Australia

To:	Senate Community Affairs References Committee
Inquiry:	Epilepsy in Australia
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Executive summary

InGeNA welcomes the opportunity to provide a submission to the Senate Inquiry into Epilepsy in Australia.

InGeNA is the genomics industry peak body. Its purpose is to make personalised healthcare accessible to all Australians by harnessing the collective skills and expertise of industry to accelerate the safe, effective and equitable adoption of genomics. Epilepsy is a clear example of why this work matters. Across Australia, epilepsy care remains too often delayed, fragmented, postcode-dependent and reactive. People with drug-resistant epilepsy, developmental and epileptic encephalopathies (DEEs), rare genetic epilepsies and complex childhood-onset epilepsies are among those most poorly served by generic pathways.

These issues are consistent with InGeNA's broader policy position: genomics is no longer experimental. Australia now needs the governance, funding, workforce, health technology assessment, data and implementation infrastructure to move genomic medicine from research and pilots into routine clinical care.

Epilepsy is an important test case for that transition. Genomic testing can shorten diagnostic delay, identify rare and syndromic epilepsies, inform clinical management, support family counselling, guide access to trials and emerging precision therapies, and reduce reliance on trial-and-error prescribing. However, genomic testing alone is not enough. It must be embedded in a nationally consistent pathway that includes genetic counselling, specialist interpretation, data reanalysis, clinical decision support, equitable access and clear links to treatment and support.

InGeNA recommends that the Australian Government establish a nationally coordinated epilepsy genomics and precision medicine pathway, supported by sustainable funding, a National Genomic Test Directory, national genomic data infrastructure, workforce capability and fit-for-purpose health technology assessment.

Summary of recommendations

InGeNA recommends that the Australian Government:

1. **Establish a national epilepsy genomics and precision medicine pathway** covering timely genomic testing, interpretation, counselling, reanalysis and referral into treatment, trials and supports.
2. **Fund a National Genomic Test Directory** that includes epilepsy and DEE indications, eligibility criteria, reimbursement status, ordering pathways and review mechanisms.
3. **Provide sustainable public funding for clinically indicated genomic testing in epilepsy**, including first-line whole exome or whole genome sequencing for suspected DEE and rare genetic epilepsy, testing for adults who missed childhood genomic diagnosis, and reanalysis as knowledge evolves.
4. **Develop fit-for-purpose HTA and reimbursement pathways** for genomic tests, digital decision-support tools, pharmacogenomics, small-cohort therapies, N-of-1 approaches and precision treatments for rare epilepsies.
5. **Support integrated precision diagnosis models such as the Australian Epilepsy Project**, where genomic testing is combined with advanced imaging, neuropsychology, digital infrastructure and specialist decision support.
6. **Build national genomic data infrastructure for epilepsy**, linking genomic, phenotypic, imaging, treatment-response, medication safety, hospital, disability and patient-reported outcome data under strong consent, privacy and governance arrangements.
7. **Strengthen genomics workforce capability** across neurology, paediatrics, general practice, pharmacy, genetic counselling, genomic pathology, bioinformatics, data science and clinical decision support.
8. **Address rural, regional, remote, First Nations and culturally diverse access gaps by design**, including tele-genetics, regional sample collection, culturally safe counselling and public funding that does not depend on private capacity to pay.
9. **Embed pharmacogenomics and precision prescribing in epilepsy care**, including evidence generation on medication response, adverse effects, genotype-informed prescribing and clinical decision support.
10. **Invest in research translation for drug-resistant epilepsy, DEEs and rare genetic epilepsies**, including targeted research calls, clinical trial matching, patient registries, natural history studies, model systems and pathways from discovery to clinical use.
11. **Ensure genomic information is used responsibly in care and support systems**, recognising that genomic results can inform care planning but should not gatekeep disability or social support.
12. **Protect public trust through strong safeguards against genetic discrimination**, privacy breaches, inequitable access and inappropriate use of genomic information.

About InGeNA

The Industry Genomics Network Alliance is a not-for-profit Australian charity and the peak body for the genomics industry. InGeNA works across industry, government, consumers, healthcare providers,

research institutions and the wider health sector to remove systemic barriers to equitable and affordable access to genomics.

InGeNA's policy work has consistently identified recurring implementation challenges in genomic medicine: fragmented access, variation between jurisdictions, complex reimbursement, insufficient workforce capacity, disconnected data systems and reliance on time-limited pilots rather than routine care.

This submission therefore focuses on the terms of reference most relevant to InGeNA's expertise: diagnosis and access to treatment, drug-resistant and rare genetic epilepsy, genomic literacy, research translation and national genomic data infrastructure.

Why epilepsy is a priority for genomics and personalised medicine

Epilepsy is not a single disease. It is a heterogeneous group of neurological conditions with different causes, seizure types, prognoses, treatment responses and disability impacts. For some people, epilepsy is controlled with medication. For others, particularly people with DEEs, rare genetic epilepsies and drug-resistant epilepsy, it is lifelong, complex, disabling and associated with significant risks including injury, cognitive impacts, developmental impairment, status epilepticus and sudden unexpected death in epilepsy.

Many families affected by rare genetic epilepsies only receive a genetic diagnosis after prolonged diagnostic uncertainty, repeated emergency presentations, intensive care admissions, specialist delays and substantial parental advocacy. Conditions such as NBEA-related epilepsy, CSNK2B-related disorder, PURA syndrome, PCDH19 epilepsy, chromosomal deletion syndromes and other rare genetic epilepsies demonstrate a repeated pattern:

- early seizures are misclassified or normalised;
- standard EEG or MRI results are treated as falsely reassuring;
- genetic testing is delayed, not offered, or only suggested after repeated escalation;
- families receive a result but limited counselling, interpretation or prognostic guidance;
- treatment remains trial-and-error despite a molecular diagnosis;
- disability and education systems do not understand the functional impact of severe genetic epilepsy;
- adults who missed genomic testing as children may remain undiagnosed.

These are not isolated clinical issues. They reflect broader structural gaps in diagnosis, treatment, research, data and national infrastructure. The Department of Health, Disability and Ageing has recognised that pharmacogenomic-guided care holds promise in epilepsy, that genomic testing can provide diagnosis and inform clinical management, and that access to genomic services varies across Australia, including disparities for Aboriginal and Torres Strait Islander people.

Term of reference (a): barriers to diagnosis and access to treatment

1. Genomic testing should be embedded in national epilepsy diagnostic pathways

The current system relies too heavily on geography, clinician awareness and family advocacy. A nationally consistent pathway should specify when genomic testing is indicated and how it is accessed, interpreted and acted on.

InGeNA recommends that genomic testing be incorporated into epilepsy pathways for:

- infants and children with recurrent, atypical, prolonged or clustered seizures;
- suspected developmental and epileptic encephalopathy;
- epilepsy with developmental delay, intellectual disability, autism, regression, movement disorder or multi-system features;
- drug-resistant epilepsy;
- suspected familial, syndromic or rare epilepsy;
- unexplained epilepsy after standard investigations;
- adults with childhood-onset or lifelong epilepsy who were never offered modern genomic testing;
- patients whose earlier testing was negative, limited or inconclusive and should be reanalysed.

The purpose should not be testing for its own sake. The pathway must connect genomic diagnosis to clinical interpretation, treatment selection, family counselling, reproductive advice, clinical trials, disability evidence and long-term review.

2. A National Genomic Test Directory is a practical access reform

InGeNA has previously called for a funded National Genomic Test Directory. Epilepsy demonstrates why it is needed. Patients and clinicians currently face inconsistent access, unclear eligibility, slow funding decisions and variation between jurisdictions.

A National Genomic Test Directory should include epilepsy-related genomic tests and should specify:

- test indications and eligibility criteria;
- whether testing is funded through MBS, state/territory pathways, public hospitals or other mechanisms;
- ordering requirements and responsible clinicians;
- expected turnaround times;
- counselling and consent requirements;
- reanalysis arrangements;
- links to clinical guidelines, patient materials and referral pathways;
- review processes to add, revise or retire tests as evidence changes.

The Directory should not be limited to cancer. Epilepsy, rare disease and paediatric neurodevelopmental conditions require equal attention if Australia is serious about mainstreaming genomics beyond isolated programs.

3. Funding should cover the whole genomic care episode

Public funding should cover more than the laboratory test. For epilepsy, the clinically useful genomic care episode includes:

- pre-test counselling and consent;
- sample collection, including regional and remote collection options;
- sequencing and analysis;
- multidisciplinary interpretation;
- written reporting that is usable by treating clinicians;
- post-test counselling;
- family implications and cascade testing where relevant;
- periodic reanalysis;
- referral to precision treatment, trials, registries and support services.

Funding only the test risks producing results that families and treating clinicians cannot use.

4. Rural and regional equity must be designed into the model

Access to genomic testing and genetic counselling should not depend on postcode, private capacity to pay, or proximity to a tertiary hospital. Australian Government material recognises variation in genomic service availability across jurisdictions and disparities in access for Aboriginal and Torres Strait Islander people.

An epilepsy genomics pathway should therefore include:

- local, regional or mailed sample collection;
- tele-genetics and telehealth genetic counselling;
- genomic decision support for regional clinicians;
- culturally safe counselling and consent processes, including for Aboriginal and Torres Strait Islander communities;
- interpreter-supported genomic counselling and patient materials;
- public funding arrangements that do not rely on private capacity to pay;
- national referral pathways that reduce cross-border variation in genomic access.

Equity cannot be left to future implementation. It must be a core design requirement for genomic medicine.

Term of reference (b): drug-resistant epilepsy and its psychosocial and economic impacts

5. Precision medicine is central to the drug-resistant epilepsy response

Drug-resistant epilepsy has disproportionate impacts on individuals, families, schools, employment, health services and disability systems. Precision medicine can support earlier and more accurate identification of patients who need escalation beyond standard medication trials. This includes:

- diagnosing genetic epilepsies with specific management implications;
- identifying patients who may benefit from precision therapies or clinical trials;
- supporting medication selection or avoidance where genotype-specific evidence exists;
- informing surgical, dietary, neuromodulation or other specialist referral decisions;
- reducing prolonged trial-and-error treatment where a more targeted approach is available.

Family-led organisations such as GETA and PURA Foundation Australia highlight this point directly: Australia has clinical and research capability, but families are still experiencing delayed genetic diagnosis, limited precision therapy access and reliance on family-led fundraising for rare epilepsy research.

6. Fit-for-purpose HTA is needed for rare epilepsy and small cohorts

Existing health technology assessment and reimbursement pathways are often poorly suited to rare genetic epilepsies. Small patient populations, rapidly evolving evidence, N-of-1 approaches and mechanism-informed therapies do not always fit conventional assessment models.

InGeNA recommends a dedicated policy pathway for evaluating genomic tests and precision therapies in rare and drug-resistant epilepsy. This should include:

- accepted methods for small-cohort evidence;
- real-world evidence generation;
- adaptive reimbursement where appropriate;
- patient-reported and carer-reported outcomes;
- long-term system cost offsets, including reduced hospitalisation, reduced ineffective treatment and reduced disability support needs;
- mechanisms to support Australian trial participation and local therapy development.

7. Pharmacogenomics should be incorporated into epilepsy medication policy

The Department of Health, Disability and Ageing acknowledges that pharmacogenomic-guided care holds promise in epilepsy because of wide variation in anti-seizure medication effectiveness and tolerability. People with epilepsy also commonly report severe side effects, medication switching, behavioural impacts, brand/formulation concerns and high treatment burden.

InGeNA recommends investment in pharmacogenomic evidence generation and implementation for epilepsy, including medication response, adverse effects, high-risk genotypes and clinical decision support.

Term of reference (c): community awareness and understanding of epilepsy and treatment options

8. Genomic literacy must extend beyond genetics services

A national genomics pathway will fail if only a small specialist workforce understands it. Epilepsy-related genomic literacy is needed across:

- general practice;
- paediatrics;
- adult neurology;
- pharmacy;
- genetic counselling;
- genomic pathology;
- bioinformatics and data science;
- digital health and clinical decision-support teams;
- policy, commissioning and reimbursement decision-makers.

Training should cover when to suspect rare or genetic epilepsy, when to refer, how to explain genomic testing, how to interpret a result in practical terms, how to use pharmacogenomic information, and how to avoid overclaiming certainty where evidence is still emerging.

9. Knowledge translation should answer the questions families actually have

International epilepsy priority-setting exercises from Canada and the United Kingdom show the value of community-led research priority setting and plain-language knowledge translation. These exercises identify personalised medicine, gene therapy, biomarkers, AI and drug-resistant epilepsy among major research priorities, reinforcing the need to translate genomics and precision medicine evidence into accessible clinical information.

Australia should fund a genomics knowledge translation program for epilepsy that provides plain-language, regularly updated information about:

- genomic testing and what results mean;
- pharmacogenomics and precision prescribing;
- variant classification, uncertainty and reclassification;
- when and how to seek genomic reanalysis;
- genotype-specific clinical trials and emerging therapies;
- reproductive and family implications of genetic epilepsy;
- responsible data sharing, privacy and consent.

This should be co-designed with people with epilepsy, families, clinicians, genetic counsellors, researchers and industry.

Term of reference (d): post-diagnosis supports including NDIS

10. A genomic diagnosis should support care planning, but not become a barrier to support

A genetic diagnosis can assist care planning, prognosis, family counselling and evidence of likely clinical trajectory. However, support eligibility must remain based on functional impact and risk, not simply on whether a person has a named gene result.

InGeNA recommends that genomic information be used appropriately to inform:

- clinical prognosis and care planning;
- developmental and cognitive risk assessment;
- family and reproductive counselling;
- eligibility for precision therapies or clinical trials;
- reanalysis and long-term review as evidence evolves;
- aggregated service planning for rare and complex epilepsy cohorts.

At the same time, people with epilepsy should not be denied supports because their genetic result is uncertain, emerging, reclassified, absent or not yet understood.

11. Rare genetic epilepsy needs navigation and multidisciplinary support

Rare genetic epilepsy care requires navigation from diagnosis to action. A genomic result should trigger clear next steps for interpretation, reanalysis, family counselling, treatment options and trial access.

InGeNA supports a multidisciplinary model in which genomic diagnosis is linked to:

- epileptology and paediatric neurology;
- clinical genetics and genetic counselling;
- genomic pathology and bioinformatics;
- pharmacy and pharmacogenomics review;
- variant reinterpretation and data reanalysis;
- disease-specific rare epilepsy groups where relevant;
- clinical trial, registry and natural history study referral.

Term of reference (e): adequacy of Commonwealth funding for research

12. Research funding should prioritise translation into routine care

InGeNA supports increased Commonwealth investment in epilepsy research, but emphasises that funding should not stop at discovery. Australia needs research translation infrastructure that moves evidence into reimbursed, equitable clinical care.

Priorities should include:

- drug-resistant epilepsy and DEEs where genomic mechanisms are implicated;
- rare genetic epilepsies;
- pharmacogenomics and medication tolerability;
- genotype-phenotype natural history studies;
- longitudinal genomic outcome datasets;
- genomic and other biomarkers for diagnosis, prognosis and treatment selection;
- AI-enabled genomic and clinical decision support;
- N-of-1 and small-cohort therapy development;
- precision therapy development and evaluation;
- clinical trial matching and recruitment infrastructure.

Research should measure patient and carer outcomes, not only academic outputs. InGeNA's broader R&D policy position is that Australia should measure improved diagnosis, treatment access, uptake of genomics and precision health tools, cost savings, clinical trials, sovereign capability, equitable access and system implementation.

13. National genomic data infrastructure is essential

Epilepsy genomics requires secure, interoperable and ethically governed data infrastructure. The Department of Health, Disability and Ageing notes Australia's obligations under the WHO Intersectoral Global Action Plan on Epilepsy and Other Neurological Disorders and the need for improved national data, timely diagnosis and workforce planning. It also notes investment in AIHW neurological conditions reporting.

InGeNA recommends building on this foundation with epilepsy-specific genomic data infrastructure that can link:

- genomic variants and reanalysis history;
- seizure type and syndrome;
- age of onset;
- EEG, MRI and neuropsychology findings;
- medication exposure, response and adverse effects;
- acute care and hospital utilisation where relevant to genomic or precision-medicine evaluation;
- surgery, dietary therapy, neuromodulation and other interventions;

- clinical trial participation;
- NDIS and support needs at aggregate, privacy-preserving levels;
- quality of life, education, work and carer outcomes.

This infrastructure should follow FAIR principles, strong consent and governance, and should be designed for clinical care, research, health service planning and industry collaboration.

14. Australia should support clinical trial matching for genetic epilepsy

Genetic diagnosis can enable trial matching and therapy development. The Australian Epilepsy Project already demonstrates how pre-phenotyped patients and linked data can make clinical trial recruitment faster, cheaper and more accurate.

A national epilepsy genomics pathway should therefore connect consenting patients to:

- clinical trial registries;
- genotype-specific studies;
- natural history studies;
- patient-reported outcome cohorts;
- international rare disease collaborations;
- emerging precision therapy programs.

Term of reference (f): other related matters

15. Pharmacogenomics and precision prescribing should be part of medicines safety

Epilepsy medicines safety should include pharmacogenomics and systematic learning about medication response, adverse effects and genotype-informed prescribing. Sodium valproate, teratogenic risk, severe adverse drug reactions, medication tolerability and brand/formulation changes all show why medication-risk information needs to be captured, communicated and used consistently.

InGeNA recommends that any national epilepsy genomics pathway include:

- pharmacogenomic evidence generation for anti-seizure medicines;
- clinical decision support for genotype-informed prescribing where evidence supports use;
- data collection on medication exposure, response and adverse outcomes;
- links between genomic results, prescribing systems and pharmacy systems;
- patient-facing information about the relevance and limits of pharmacogenomic results;
- reproductive and family counselling where genomic or medication-risk information has implications for relatives or future pregnancies.

This should support informed choice and safe care without reducing access to effective treatment for people who need it.

16. Genetic discrimination protections are necessary for public trust

Genomic medicine depends on public trust. People may avoid testing if they fear that a genetic result could affect insurance, employment, family members or future opportunities. InGeNA has supported stronger protections against genetic discrimination and recommends that epilepsy genomics implementation be accompanied by clear safeguards for privacy, consent, appropriate use and non-discrimination.

17. The Senate inquiry should recommend genomics implementation machinery, not only principles

International precedents show the importance of measurable targets, implementation accountability and public reporting. InGeNA recommends that the Committee's report include genomics-specific implementation machinery, including:

- responsible Commonwealth and state/territory agencies;
- implementation timeframes;
- indicators for genomic testing access, turnaround time and equity;
- indicators for genetic counselling availability and workforce capacity;
- reporting on variant reanalysis and reinterpretation pathways;
- data interoperability and clinical decision-support milestones;
- pharmacogenomics uptake where evidence supports clinical use;
- clinical trial and registry matching measures;
- mechanisms for consumer, clinician, research and industry input;
- periodic review as genomic evidence evolves.

Proposed national implementation model

InGeNA proposes that the Commonwealth, in partnership with states and territories, consumers, clinicians, researchers, community organisations and industry, establish a national epilepsy genomics and precision medicine implementation program with five linked components.

Component 1: National pathway

A consensus clinical pathway for suspected rare, complex, drug-resistant and developmental epilepsy, including testing indications, referral criteria, counselling, interpretation, reanalysis and escalation to specialist care.

Component 2: Funding and test directory

Public funding for clinically indicated testing and counselling, supported by a National Genomic Test Directory that includes epilepsy indications and is reviewed regularly.

Component 3: Integrated care and decision support

Support for integrated diagnostic models, including AEP-like approaches, that combine genomics with imaging, neuropsychology, digital tools and specialist interpretation.

Component 4: Data and research translation

A national genomic epilepsy data and trial-matching infrastructure, built on consent, privacy, interoperability and FAIR principles, supporting clinical care, research, health service planning and precision therapy development.

Component 5: Genomic workforce, equity and trust

Investment in genomic literacy, genetic counselling capacity, tele-genetics, regional genomic access, culturally safe genomic counselling, anti-discrimination protections and public trust.

Conclusion

People with epilepsy and their families are carrying the cost of system fragmentation. This is most visible in rare genetic epilepsies, DEEs and drug-resistant epilepsy, where delayed diagnosis, trial-and-error treatment, limited specialist access, inadequate data and weak support pathways compound harm across the lifespan.

Australia has the clinical, research and industry capability to do better. The missing element is not scientific possibility. It is implementation: national pathways, sustainable funding, data infrastructure, workforce capability, equitable access and accountability.

InGeNA urges the Committee to recommend a national epilepsy genomics and precision medicine pathway as a practical, high-impact reform that can shorten diagnostic delay, improve treatment selection, support research translation and deliver more equitable care for Australians living with epilepsy.