DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHIES

Collective Voices from the DEE Community

134 families share what life is actually like — and what must change.

A submission of lived experience to the Senate Inquiry into Developmental and Epileptic Encephalopathies.

FACES OF THE DEE COMMUNITY · SHARED WITH FAMILY PERMISSION

> *"We are families who have become experts — in science, in policy, in advocacy, and in one another. The system was not designed for us. It can be."*
>
> SCN2A AUSTRALIA

THE COMMUNITY IN NUMBERS

134 families. One message: this system is not built for us.

Families living with Developmental and Epileptic Encephalopathies (DEE) responded to an open call from SCN2A Australia and consented to share their experience with the Senate Inquiry. Their answers reveal a community navigating drug-resistant epilepsy, fragmented care, and an NDIS that frequently does not recognise the complexity it sees in front of it.

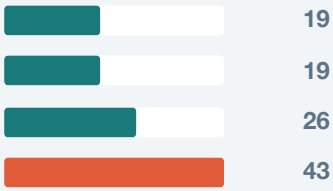
134

Family responses — each consenting to share their lived experience with the Senate Inquiry

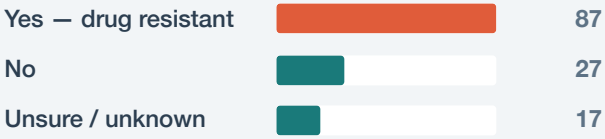
76%

Report drug-resistant epilepsy†

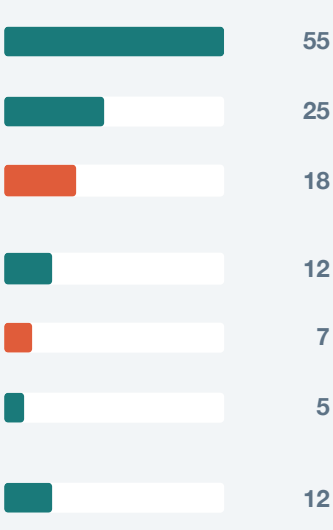
Age of the person living with DEE (n=107 reporting age)



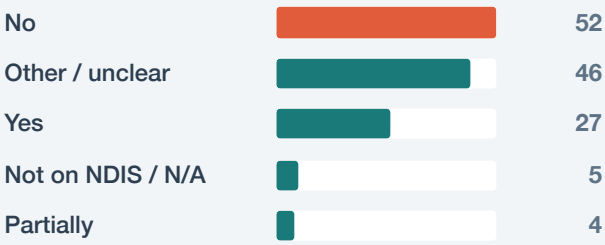
Drug-resistance status reported (n=131)



Reported diagnoses — grouped (n=134)



Does your NDIS plan respond to the complexity of your child's conditions?



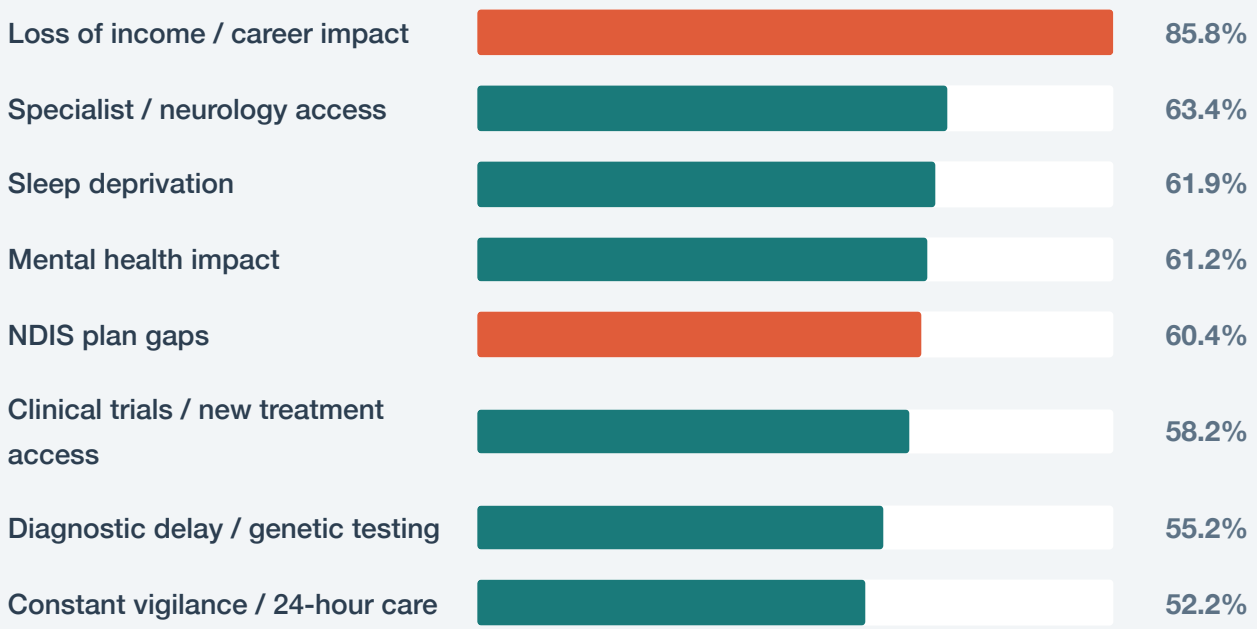
† Drug-resistance share calculated across the 114 respondents giving a clear yes/no. ‡ Includes SYNGAP1, PCDH19, KCNQ2, STXBP1, Angelman, TSC and others — each named by 1–3 families.

EIGHT THEMES — REPEATED ACROSS 134 VOICES

The same problems are showing up in every postcode.

WHAT RISES TO THE TOP

Read as a whole, the responses describe a system that requires families to be **specialists, coordinators, and advocates** simultaneously — while also providing 24-hour care, managing medications, and absorbing the financial loss that comes with reduced work. Drug-resistance is not the exception; it is the norm.



About this graph. Open-text answers were scanned for recurring themes. The figures show the share of respondents whose answers touched on each theme — drawn from comments on diagnosis, daily life, access barriers, family impact, research, and the change they want to see.

> *"Everything from doctor understanding, Centrelink rejections, NDIS rejections and general struggle to make do and get by."*
>
> — FAMILY RESPONDENT

4 in 5
families describe a direct hit to income and career

3 in 5
describe sleep loss, mental-health impact, and NDIS plan gaps

1 in 2
describe constant vigilance — 24-hour care with no respite

IN THEIR OWN WORDS

What the day-to-day actually looks like.

85.8% of responses

Loss of income and career

> *"I had to give up my professional career as a doctor."*

> *"Down to single income household after needing to resign to be full-time carer."*

> *"I can't work more than two days a week when I have support or someone that can ensure his safety."*

> *"My family members also had to take time off work to take me to appointments."*

61.9% of responses

Sleep deprivation

> *"I don't sleep through more than one to two nights per week due to her sleep condition."*

> *"My husband and I can't sleep together anymore — I sleep with our daughter to monitor her at night."*

> *"One parent will sleep in our son's room every night to reduce the risk of SUDEP."*

> *"Lack of sleep — waking several times a night due to seizures."*

61.2% of responses

Mental health

> *"My mental health is the worst it has ever been. I watch my daughter have seizures daily."*

> *"I have PTSD from traumatic medical episodes of epilepsy."*

> *"Both parents have suffered grief, depression and anxiety."*

> *"I try so hard to stay positive for my son but cry a lot when he's not around."*

52.2% of responses

Constant vigilance

> *"There is no switching off. There is no family member or friend we could ask to look after him."*

> *"Our daughter lives 'under surveillance' — that's how I explain it to people."*

> *"We are exhausted from being constantly vigilant to prevent, detect and respond to seizures."*

> *"The acute and chronic nature of this condition means you have to be hypervigilant at all times."*

> ***"Stolen our whole lives — socially, economically, mental health, sleep — as it requires full-time care."***
>
> — FAMILY RESPONDENT

WHERE THE SYSTEM STOPS WORKING

Specialists, NDIS, diagnosis, and research — the four breakpoints.

63.4% of responses

Lack of access to a specialist

> *"She was diagnosed by a hospital neurologist in December — unable to get an appointment to see a specialist until April."*

> *"Living in regional NSW it is hard to see a specialist with our closest children's hospital three hours away."*

> *"We are living in Mackay and need to travel to Townsville to see a specialist."*

> *"Wait times for neurologist are insane, and the cost of consultations is very high."*

60.4% of responses

NDIS plan gaps

> *"NDIS — been told epilepsy isn't part of it, denied everything that has to do with epilepsy."*

> *"A NDIS that truly sees my child and does not treat her like she's a red line in the budget."*

> *"NDIS has been so difficult to deal with — to the point of us considering walking away and taking over all care."*

> *"NDIS needs to recognise that people with uncontrollable epilepsy need around-the-clock care to keep them safe."*

55.2% of responses

Diagnostic delay & genetic testing

> *"It is estimated only 2% of the whole RNU 4-2 population in Australia has received a diagnosis — due to difficulty accessing the appropriate test."*

> *"Told because of her age that genetic testing would be too expensive."*

> *"Took a long time for diagnosis. Long waitlist for MRI, EEGs etc."*

> *"Children like my son are being missed due to difficulty accessing an accurate diagnosis."*

58.2% of responses

Lack of access to clinical trials and treatments

> *"Australia has nothing really to offer — it seems behind the times. We are looking into clinical trials overseas."*

> *"A trial we are seeking to access has been delayed by months compared to when it started in the US."*

> *"There is no CNC funding to allow for a clinic, meaning access to clinical trials will only happen if we raise money through a charity."*

> *"I personally spent $20,000 on a medication out of my own super to try and help my child."*

THE SINGLE MOST IMPORTANT CHANGE

What families said, when asked for one thing.

Q: *What is the single most important change needed to improve epilepsy care for families like yours?* — A clear cluster of asks emerged across the 134 consenting respondents.

> *"Recognise refractory epilepsies and DEEs as a disability — that will take a load off our parental shoulders."*
>
> — FAMILY RESPONDENT

1. Recognise DEE as a disability

NDIS, Centrelink and education systems must treat drug-resistant DEE as the lifelong, complex disability it is — not as a medical condition outside their remit.

2. Genetic diagnosis for all

Whole genome sequencing as standard, not as a barrier. A diagnosis unlocks trials, targeted therapy, and a community.

3. Specialist multidisciplinary clinics

Coordinated DEE clinics with clinical nurse consultant (CNC) capacity, accessible from regional Australia, with paediatric-to-adult transition.

4. Trials and treatments in Australia

Funded research infrastructure so families do not have to fundraise — or fly overseas — to access emerging gene therapies and trials.

5. Funded respite and trained support workers

Carers competent with seizure first aid and midazolam. Reliable in-home support so primary carers can sleep, work, and survive.

6. Mental health support for the whole family

Psychology, peer support and trauma-aware care for parents and siblings — not just for the person with epilepsy.

These six asks are derived from clustering the open-text responses to the "single most important change" question. They are the consistent through-line: an NDIS that responds to complexity, faster pathways to a genetic diagnosis, coordinated specialist care close to home, Australian access to trials, funded support that allows carers to remain in work, and recognition that DEE is a whole-of-family condition.

METHODOLOGY, CONSENT, AND CONTACT

How this evidence was gathered.

Source

134 responses to an online questionnaire circulated by SCN2A Australia to families affected by Developmental and Epileptic Encephalopathies and related rare epilepsies, in response to the Senate Inquiry. All respondents consented to share their experience for advocacy and reform.

Consent

This report represents only the 134 respondents who explicitly consented to their de-identified experience being used to support advocacy and reform. All direct quotes are reproduced verbatim, with minor edits for clarity only.

Quantitative analysis

- Age, diagnosis category, drug-resistance status and NDIS-adequacy responses were coded from structured fields.
- The diagnosis grouping reflects the most-named gene or syndrome in each response. Many families named more than one.
- “Drug-resistant” share is reported as a proportion of respondents giving a definitive yes or no.

Thematic analysis

- Eight themes were identified across open-text answers covering daily life, access barriers, family impact, research, and asked-for change.
- Theme prevalence is the share of respondents whose answers contained one or more keywords associated with that theme.
- Quotes were selected for clarity and representativeness, not extremity.

Families became the experts because the system did not. They are ready to be partners — not petitioners.

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