Submission to the Senate Inquiry into Epilepsy Services and Support in Australia

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Introduction

I am the mother of a young child living with epilepsy, following a prolonged and traumatic journey to diagnosis. We live in Orange, New South Wales, and our experience highlights the significant barriers faced by regional families in accessing timely diagnosis, appropriate treatment, and ongoing support.

My daughter is a happy, intelligent, and developmentally typical little girl. However, from early 2023, she began experiencing recurrent seizure activity that was repeatedly dismissed as "normal" febrile responses. Despite multiple presentations to our local emergency department, no meaningful investigation into epilepsy was undertaken until a life-threatening event forced escalation.

She has since been diagnosed with epilepsy and, more recently, a rare genetic condition involving a mutation in the **NBEA gene**. This diagnosis has brought with it not only answers, but also significant uncertainty due to the limited understanding of this condition.

Our journey to diagnosis was not delayed by complexity, but by repeated dismissal, systemic gaps in regional healthcare, and a lack of recognition of parental concerns.

Timeline and Barriers to Diagnosis

Between January 2023 and early 2024, my daughter experienced multiple seizure-like episodes, including non-responsiveness, convulsions, and prolonged post-ictal symptoms. On each occasion, we presented to Orange Health Service Emergency Department.

Across these presentations:

- Episodes were consistently attributed to "febrile seizures," even when seizures occurred **without fever at onset**
- A normal EEG was used to reassure clinicians and avoid further investigation
- No long-term neurological plan or referral pathway was established
- My concerns regarding potential epilepsy, including SCN1A-related conditions, were dismissed as unlikely due to their rarity

At one point, documentation explicitly noted the absence of an epilepsy diagnosis, despite a clear and escalating pattern of events.

Over time, the seizures increased in frequency and severity. Despite this, care remained reactive rather than preventative. Opportunities for early intervention were repeatedly missed.

Critical Incident and Escalation of Care

In April 2024, my daughter experienced a prolonged seizure lasting over 90 minutes. She did not respond to initial emergency medication and required intubation.

Due to the lack of a paediatric intensive care unit in Orange, she was stabilised in an adult ICU before being transferred to Sydney under emergency conditions.

This was the most traumatic experience of our lives. We were told there was no way of knowing what her neurological outcome would be.

Upon arrival at a tertiary hospital, a senior clinician reviewed her history and advised:

“This should not have gotten to this point.”

We were further told that had we presented multiple times to a metropolitan hospital such as Westmead or Randwick, a full investigation would likely have occurred after the third or fourth seizure not after the 13th life threatening seizure.

This acknowledgment was both validating and deeply distressing. It confirmed that geography, not medical complexity, had influenced the standard of care my daughter received.

Systemic Failures Identified

Our experience highlights several critical failures within the current system:

1. Delayed Diagnosis

Repeated seizure presentations were misclassified as febrile events, delaying appropriate neurological investigation and treatment.

2. Dismissal of Parental Concerns

Despite consistently raising concerns and independently researching potential causes, my input was frequently disregarded. This contributed directly to delays in escalation.

3. Over-reliance on Normal EEG Results

A single normal EEG was used to effectively rule out epilepsy, despite ongoing clinical symptoms.

4. Lack of Specialist Access in Regional Areas

There was no direct or timely access to paediatric neurology services. Referrals were inconsistent and often deprioritised.

5. Reactive Rather Than Preventative Care

Treatment only escalated after a life-threatening event, rather than at earlier stages where intervention may have prevented harm.

6. Geographic Inequality

We were explicitly told that care would have differed had we lived in Sydney. This highlights a clear disparity between metropolitan and regional healthcare standards.

Challenges Following Diagnosis and Rare Genetic Epilepsy

Following our daughter's diagnosis and engagement with a specialist neurology team, her seizures are now largely controlled with medication.

More recently, she has undergone genetic testing and has been identified as having a mutation in the **NBEA gene**, a rare and emerging genetic condition associated with epilepsy and neurodevelopmental differences.

This diagnosis has introduced a new layer of complexity and uncertainty for our family.

Limited Information and Clinical Guidance

NBEA-related epilepsy is not widely understood. There is:

- Limited clinical information available
- No clear long-term prognosis
- Minimal guidance on what to expect as she grows

We are navigating a condition where even specialists acknowledge that there are still many unknowns.

Uncertainty Around Development and Future Outcomes

While our daughter is currently developing well, we do not know:

- Whether her epilepsy will remain controlled
- If she may experience developmental, behavioural, or cognitive impacts over time

- What supports she may need in the future

This uncertainty places an ongoing emotional strain on our family, as we are required to plan for possibilities without clear direction.

Lack of Structured Support for Rare Epilepsies

Despite having a confirmed genetic diagnosis:

- There is no clear support pathway specific to her condition
- We have not been provided with tailored resources or long-term planning frameworks
- Much of our understanding continues to come from our own research and advocacy

Cost of Treatment

Her medication (Clobazam) is compounded and not subsidised under the PBS in this form, meaning we pay full cost out-of-pocket.

Lack of Recognition Under NDIS

We applied for funding for nocturnal seizure monitoring equipment, a critical safety measure, and were denied on the basis that our daughter was not considered "disabled enough." This resulted in over $1000 out of pocket to purchase the nocturnal monitor suggested by the Epilepsy Action nurse who visited us at Randwick during Safiya's ICU stay.

This is particularly concerning given:

- The known risks associated with epilepsy, including seizures during sleep
- The unpredictable nature of genetic epilepsies
- The preventative role that monitoring equipment can play in safeguarding a child's life

Ongoing Emotional and Practical Burden

Daily medication administration remains distressing for a young child and emotionally taxing for our family.

We are not only managing a diagnosed condition, but also the unknown trajectory of a rare genetic disorder that lacks clear pathways for support.

Rural and Regional Impact

Living in a regional area significantly impacted our experience:

- No access to paediatric ICU care locally
- Delayed escalation to specialist services
- Reliance on emergency transport to metropolitan hospitals
- Inconsistent continuity of care
- Reduced access to up-to-date clinical knowledge and rare condition awareness

Our experience demonstrates that postcode should not determine the quality or timeliness of medical care.

Recommendations

Based on our experience, I urge the inquiry to consider the following:

1. **Mandatory escalation protocols** after repeated seizure presentations, regardless of presumed cause
2. **Improved education and training** for emergency clinicians on epilepsy and limitations of EEG testing
3. **Greater access to paediatric neurology services** in regional areas, including telehealth pathways
4. **Recognition of parental concerns** as valid clinical input in paediatric cases
5. **Funding support for seizure monitoring equipment** through NDIS or alternative programs
6. **PBS coverage for compounded anti-epileptic medications**
7. **Clearer diagnostic pathways for rare and genetic epilepsies**
8. **Equitable standards of care between regional and metropolitan hospitals**

Conclusion

We are incredibly grateful that our daughter survived her experience and is now receiving appropriate care.

However, it should not have taken a life-threatening event for her condition to be taken seriously.

There were multiple opportunities for intervention that were missed. Our experience reflects a broader systemic issue affecting families, particularly in regional Australia.

For families like ours, a diagnosis does not always bring clarity, sometimes it brings more questions than answers. The system must be equipped to support not only known conditions, but also those that are still being understood.

I share our story in the hope that meaningful changes can be made so that other families are not forced to endure the same trauma before receiving the care they need.