Tuberous Sclerosis Australia

ABN 20 681 174 734
Incorporation no. Y07116-42
Registered Charity CC25313
CFN 13968

About this submission

TSA welcomes the opportunity to contribute to the Senate Community Affairs References Committee Inquiry into Epilepsy in Australia.

This submission, made on behalf of the community we represent, focuses on the unique challenges experienced by people in Australia living with TSC-associated epilepsy and highlights opportunities for earlier intervention, improved access to specialised care and stronger support systems to improve long-term outcomes.

About Tuberous Sclerosis Australia (TSA)

TSA (Tuberous Sclerosis Australia) is the national patient support organisation for individuals and families affected by Tuberous Sclerosis Complex (TSC). TSA provides information and support to people affected by TSC and works with clinicians, researchers and other relevant parties to advocate for improved health and social outcomes for people living with TSC across Australia.

About Tuberous Sclerosis Complex (TSC)

TSC (Tuberous Sclerosis Complex) is a rare, genetic, multi-system condition caused by mutations in the *TSC1* or *TSC2* genes. These mutations lead to over-activation of the mTOR pathway, causing benign tumours and abnormal tissue growth in multiple organs, including the brain, kidneys, heart, lungs, eyes and skin.

TSC is a lifelong condition requiring co-ordinated multidisciplinary care across paediatric and adult health systems. Epilepsy is the most common and often the most severe clinical feature of TSC.

Individuals with TSC commonly require care from neurologists, epileptologists, nephrologists, geneticists, psychiatrists, psychologists, allied health professionals and disability services throughout their lives.

TSC affects approximately 1 in 6,000–10,000 people and is one of the more common rare diseases associated with epilepsy. It has the same prevalence as motor neurone disease (MND) and is one of the leading genetic causes of epilepsy and autism spectrum disorder[1]. Approx. two-thirds of people diagnosed with TSC have no prior family history.

TSC-associated epilepsy

Epilepsy is the most common and often most severe clinical feature of TSC, affecting approximately 60–90% of individuals[2]. TSC-associated epilepsy is characterised by:

- early onset - frequently in infancy
- multiple seizure types, including infantile spasms

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- high rates of drug-resistant epilepsy
- frequent hospitalisation and emergency presentations
- strong association with developmental and neurocognitive impairment.

Importantly, epilepsy in TSC is not simply a seizure disorder.

Many children with TSC experience features consistent with developmental and epileptic encephalopathy (DEE), where epileptic activity itself contributes to developmental slowing or regression[3].

Seizure activity during infancy and early childhood can directly disrupt brain development and significantly affect long-term cognitive, behavioural and psychosocial outcomes.

Children with DEE may experience:

- developmental regression
- intellectual disability
- communication impairment
- behavioural and psychiatric challenges
- autism spectrum disorder
- loss of developmental milestones.

Early recognition and effective seizure control can improve developmental trajectories and reduce lifelong disability.

The burden of TSC-associated epilepsy therefore extends well beyond seizures and significantly impacts families, carers, education systems, disability services and the broader economy[4].

Lilly

From just 9 weeks of age, Lilly began experiencing seizures. Her parents, Dave and Erin were told by doctors that her outlook was uncertain and that her long-term development may be severely affected. Over the years, the family has trialled numerous medications in an attempt to control Lilly's seizures, but the journey has been relentless and exhausting. Lilly, who is now 12 years old, has experienced frequent seizures, sometimes up to 10 per day, including episodes at preschool and school. She has suffered injuries from seizure-related falls, including broken bones, requiring repeated emergency hospital visits.

Key issues and recommendations

There have been significant advances in TSC-related epilepsy care globally in the past two decades and there are many patients in Australia who have benefitted from these advances. However, too often TSA's Community still experiences disparity with regards to epilepsy care in Australia leading to unnecessarily poor patient outcomes.

1. Infantile spasms need to be recognised as a neurological emergency

Infantile spasms are frequently the first presenting seizure type in TSC. They require urgent investigation, diagnosis and treatment. These spasms are often an indicator of TSC but are often missed or misdiagnosed.

Infantile spasms may initially be subtle and can be mistaken for reflux, colic or normal infant behaviour, often contributing to delayed diagnosis[5].

Delayed treatment is associated with:

- worsening epilepsy
- developmental regression
- poorer long-term cognitive outcomes
- increased lifelong disability support needs.

This issue is particularly significant in regional and rural Australia, where access to paediatric neurologists and urgent VEEG assessment is often very limited.

Recommendations

- Increase awareness and education of infantile spasms among frontline healthcare providers to support earlier recognition and urgent treatment initiation.
- Develop nationally consistent, time-critical referral and escalation pathways to ensure infants and children with suspected spasms or severe epilepsy syndromes receive urgent neurological assessment and rapid access to specialist paediatric epilepsy services.
- Improve equitable access to urgent paediatric video EEG (VEEG) services including inpatient and ambulatory VEEG capability, with timely specialist interpretation to reduce delays in diagnosis and treatment.
- Support sustained national public awareness and caregiver education initiatives to improve early recognition of infantile spasms and other seizure disorders.

2. Early seizure treatment and VEEG surveillance improve outcomes

There is strong evidence that early and aggressive treatment of seizures improves developmental and neurocognitive outcomes in TSC[6].

Emerging evidence also supports VEEG surveillance and treatment of epileptiform activity before clinical seizures appear[7].

Regular VEEG monitoring and early intervention may:

- delay or reduce epilepsy onset
- reduce seizure severity
- improve neurodevelopmental outcomes
- reduce the risk of drug-resistant epilepsy.

However, implementation requires:

- regular VEEG monitoring in infancy
- timely specialist interpretation
- coordinated follow-up
- equitable access to paediatric neurology services.

Access to VEEG services remains inconsistent across Australia, particularly for families living outside metropolitan centres.

Katalina

From soon after her birth, Katalina began experiencing seizures, marking the beginning of a long and difficult journey to manage her condition. Her parents, Boris and Kirsten, faced ongoing uncertainty as they searched for answers and effective treatment options.

Part of this struggle involved accessing specialised diagnostic care. At the time, the family was devastated to learn that overnight video EEG monitoring, an important tool used to assess and manage epilepsy, was not available in Perth - thankfully, it has since been reinstated.

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Recommendations

- Recognise early seizure treatment in TSC as time-critical within national epilepsy policy and clinical guidance.
- Improve access to rapid specialist assessment and treatment initiation for infants with suspected TSC-associated epilepsy.
- Support nationally coordinated clinical pathways for early diagnosis, VEEG surveillance and treatment of TSC-associated epilepsy.
- Integrate VEEG-based surveillance into standard TSC care pathways.
- Expand telehealth-supported neurology and epilepsy services, particularly for regional and rural families.
- Improve Medicare and hospital funding arrangements to support repeat VEEG monitoring where clinically indicated.
- Improve equitable access to specialist epilepsy services regardless of postcode.

3. Access to precision therapies and comprehensive epilepsy care

TSC is caused by dysregulation of the mTOR pathway. mTOR inhibitor therapies, including everolimus, directly target the underlying disease mechanism[8].

These therapies:

- reduce seizure frequency
- are effective in drug-resistant epilepsy
- treat multiple manifestations of TSC
- represent an important example of precision medicine in epilepsy care.

Despite the advances in these treatments, many people with TSC continue to require other forms of comprehensive epilepsy management including:

- anti-seizure medications
- ketogenic diet therapy
- epilepsy surgery
- vagus nerve stimulation (VNS)
- neuropsychology and behavioural supports.

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Access to these treatments, which are well proven to be beneficial to patients living with TSC-related epilepsy remains widely inconsistent across Australia, even in major cities and particularly in regional and remote areas. Patients report different experiences in being made aware of such therapies and being offered them as options. For example, patients in Perth and more widely in WA frequently report a lack of information and/or access to epilepsy surgery particularly for patients with drug-resistant epilepsy where timely assessment for epilepsy surgery is recognised as a standard component of evidence-based care[9]. Such surgery conducted in the major children's hospitals in Melbourne, Sydney and Brisbane, has proven highly effective as a management strategy for many patients living with TSC-related epilepsy.

Recommendations

- Ensure continued equitable PBS access to mTOR inhibitor therapies.
- Improve access to the full range of evidence-based epilepsy treatments across the country, including timely referral to established comprehensive epilepsy and epilepsy surgery centres where required, regardless of the patient's state or territory of residence, with appropriate support for interstate assessment, treatment and follow-up.
- Increase travel and accommodation assistance for patients and families who are required to access specialised epilepsy services interstate or far from home, to reduce financial and geographic barriers to timely care.
- Support the development and funding of advanced epilepsy surgery programs including access to contemporary diagnostic and surgical techniques such as stereoelectroencephalography (stereo EEG) and other minimally invasive epilepsy surgery approaches.
- Expand regional outreach and telehealth services.
- Support multidisciplinary epilepsy care models to deliver comprehensive, patient-centred care across diagnosis, treatment, surgery evaluation, rehabilitation, psychosocial support and long term follow-up.

Greta

When Greta was three years old, her neurologist recommended brain surgery after it became clear that her seizures could not be controlled. Her parents felt they had little choice, knowing that without intervention, Greta's future was uncertain.

The surgery was complex, lasting more than 12 hours, but it marked a turning point. Although Greta continued to experience seizures immediately afterwards, they became less frequent and less severe. Most significantly, her development rapidly improved, particularly her language and motor skills.

Several months later, Greta's seizures stopped completely. She is now seizure-free and no longer requires medication. Her family describes the surgery as life-changing.

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4. Regional inequity and access barriers

As already discussed, there are wide disparities in the standard and options of epilepsy care across the country. Within our TSC Community we are aware of a number of families who have chosen to fly their sick children interstate to get access to alternative care. In particular, patients and families living in regional, rural and remote Australia often face substantial barriers to epilepsy care, including:

- limited access to paediatric neurologists
- workforce shortages
- delayed VEEG access
- reduced access to ketogenic diet programs and epilepsy surgery assessment
- significant travel and financial burdens.

These inequities create what is effectively a postcode lottery for epilepsy care and contribute to delayed treatment and poorer outcomes for patients.

Recommendations

- Expand regional neurology and epilepsy services to reduce geographic inequity and reliance on metropolitan centres and improved access to care closer to home.
- Improve access to VEEG monitoring across regional and remote Australia.
- Support structured regional shared-care models between local healthcare providers and tertiary epilepsy centres to enable coordinated management.
- Increase investment in telehealth, outreach programs and visiting specialist programs to improve access to epilepsy expertise.

Holly

Holly's mum explains that living in a rural area has been one of the greatest challenges of raising a child with TSC. Access to emergency ambulance services can be limited, with ambulances often attending emergencies in neighbouring towns when urgently needed. During Christmas, the local hospital, where Holly and her condition are well known, closes for several weeks. Any medical emergency during this time requires the family to travel a considerable distance to another hospital, where the family must explain Holly's complex condition and emergency management plan to unfamiliar emergency staff.

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5. Families and carers require stronger support, including within the NDIS

TSC-associated epilepsy places substantial, on-going demands on families and carers.[10]

Families commonly have to manage:

- frequent hospitalisations and specialist appointments
- complex medication regimens
- constant supervision due to seizure risk
- behavioural and developmental challenges
- impacts on employment, education and mental health (both the patients and their own).

Parents and carers frequently have to reduce work hours or leave the workforce entirely because of the pressures of their caregiving responsibilities. Burnout and significant mental health issues in carers is relatively common and feelings of anxiety and isolation are part of everyday life.

Families also report significant difficulties navigating the NDIS for financial supports and report inconsistent decision-making (both for individual families and between different families) and limited understanding of rare epilepsies within the system.

Recommendations

- Ensure access to:
  - seizure monitoring devices
  - respite care
  - care coordination
  - psychosocial support.
- Increase support for carers managing high-complexity epilepsy.
- Improve recognition of rare epilepsies within the NDIS.
- Improve rare disease epilepsy education for NDIS planners and assessors.

Rachel

Due to her daughter's complex needs, Rachel stopped working when she was five years old. Rachel found it increasingly difficult to balance employment with the demands of managing her daughter's TSC, epilepsy and care needs

6. Research investment and Centres of Expertise can improve outcomes

TSC is one example of how sustained biomedical research investment can improve outcomes for people living with epilepsy. International and Australian research collaborations have led to:

- precision therapies targeting the mTOR pathway
- improved understanding of epileptogenesis
- earlier intervention strategies
- advances in epilepsy surgery
- improved developmental outcomes.

Due to the complexity of TSC and TSC-related epilepsy, outcomes are improved when care is co-ordinated through specialised multidisciplinary Centres of Expertise with experience in rare epilepsies and TSC. There is also strong evidence of a need to support the transition from paediatric to adult care otherwise patients can become lost to the system and fail to undergo the lifelong surveillance and screening that is necessary for their condition.

Australian specialist clinics and research teams have contributed significantly to international advances in TSC-related epilepsy care. However, access to specialised expertise remains inconsistent nationally.

Debbie

Our TSC Community is so wonderful because our stories are so varied and different. TSC affects everyone differently. And, since our daughter was born 20 years ago, there have been amazing advances in diagnosis, treatment and surveillance, which help improve the outcomes for people living with TSC.

Recommendations

- Support the development and sustainable funding of nationally co-ordinated Centres of Expertise in every major city in Australia with defined standards for multidisciplinary care.
- Strengthen and fund national collaborative clinical networks to support co-ordinated care and equitable access to specialist expertise across jurisdictions.
- Increase dedicated Commonwealth investment in epilepsy research through MRFF and NHMRC including support for translational research.
- Invest in national epilepsy registries and co-ordinated research infrastructure to enable benchmarking, long-term outcome monitoring and participation in multicentre research initiatives.
- Strengthen transition pathways between paediatric and adult services through structured transition programs and co-ordinated multidisciplinary support to reduce gaps in care during adolescence and early adulthood.

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Summary

The TSC Community represents a distinct and important subgroup of patients with epilepsy in Australia, characterised by:

- early-onset and often severe epilepsy
- developmental and epileptic encephalopathy
- high rates of drug-resistant epilepsy
- significant neurodevelopmental and psychosocial impacts
- complex lifelong care needs.

There are clear opportunities to improve patient outcomes through:

- earlier recognition and intervention
- equitable access to specialised diagnostics and therapies
- co-ordinated multidisciplinary care
- stronger support for families and carers
- sustained investment in research and Centres of Expertise.

In this submission we have made a number of recommendations with respect to:

1. The need for infantile spasms to be recognised as a neurological emergency

2. Improving outcomes through early seizure treatment and VEEG

3. Enabling equitable access to precision therapies and comprehensive epilepsy care

4. Reducing regional inequity and access barriers

5. Providing families and carers with stronger support, including within the NDIS

6. Investing in research and Centres of Expertise to improve outcomes

We believe there are significant opportunities to reduce long-term disability, improve participation and quality of life, and reduce costs across health, disability and social systems.

TSA would be pleased to provide further information.

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