Case 1:
54 y female with Tuberous Sclerosis

Michael K. Sowell, MD, FAHS
Professor of Neurology
Associate in Pediatrics
University of Louisville School of Medicine

UofL SCHOOL OF MEDICINE | UofL Health | Brain & Spine Institute

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54 yo female follow-up Tuberous Sclerosis; episodic and chronic migraine

FU episodic and chronic migraine

- Past Medical History includes:

- **Neurological**
- Tuberous Sclerosis
- Intellectual and Developmental Disability
- Subependymal Giant Cell Astrocytoma (frontal horn of the right lateral ventricle)
- Lumbar radiculopathy
- Paroxysmal Nonepileptic Spells

- **Behavioral Health**
- Anxiety/depression/PTSD
- PNES

- **Pulmonary**
- Pulmonary lymphangioleiomyomatosis (LAM)/chronic lung disease/oxygen dependent
- COPD/Chronic interstitial lung disease
- OSAS

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Tuberous Sclerosis (continued)

- **Cardiovascular**
- HTN
- HLD

- **Gastroenterology**
- GERD/GI hemorrhage
- Irritable bowel syndrome

- **Renal**
- stage 3a chronic kidney disease

- **Endocrinology and Metabolism**
- Obesity
- PCOS
- Type II Diabetes mellitus

- **Miscellaneous**
- Osteoarthritis

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TSC diagnostic criteria

The clinical and genetic diagnostic criteria of 2021 are summarized below:

MAJOR FEATURES	MINOR FEATURES
Hypomelanotic macules (≥3; at least 5mm diameter)	"Confetti" skin lesions
Angiofibroma (≥3) or fibrous cephalic plaque	Dental enamel pits (>3)
Ungual fibromas (≥2)	Intraoral fibromas (≥2)
Shagreen patch	Retinal achromatic patch
Multiple retinal hamartomas	Multiple renal cysts
Multiple cortical tubers and/or radial migration lines	Nonrenal hamartomas
Subependymal nodule (≥2)	Sclerotic bone lesions
Subependymal giant cell astrocytoma	
Cardiac rhabdomyoma	
Lymphangioleiomyomatosis (LAM)*	
Angiomyolipomas (>2)*	

Definite TSC: 2 major features or 1 major feature with 2 minor features.
Possible TSC: either 1 major feature or ≥2 minor features.
* a combination of the 2 Major clinical features LAM and angiomyolipomas without other features does not meet criteria for a definite diagnosis.
Genetic diagnosis: A pathogenic variant in *TSC1* or *TSC2* is diagnostic for TSC. Most TSC-causing variants are sequence variants that clearly prevent TSC1 or TSC2 protein production. Some variants compatible with protein production (e.g., some missense changes) are well established as disease-causing. Other variant types should be considered with caution.

WHAT IS TSC?
GENETICS
CLINICAL MANIFESTATIONS +
PROGNOSIS
DIAGNOSTIC CRITERIA
SURVEILLANCE & TREATMENT

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Adult support

Being diagnosed with tuberous sclerosis complex (TSC) can cause significant challenges within your adult life regardless of when you first received your diagnosis. The TSC Alliance is here to help.

From the stress of learning a new diagnosis to potentially processing the emotions of an offspring with this diagnosis to learning how to navigate the medical complexity of routine follow-up surveillance and management in the adult healthcare sector, all while maintaining other responsibilities, can be overwhelming.

In 2012, the TSC Alliance launched our Adult Initiative to prioritize the needs of adults with TSC, invest in research to understand how TSC changes in adulthood, and develop a support network and resources for both young adults and adults. You can find many of these resources in the Navigating the transition years of TSC and Navigating the adult years of TSC guides.

Below you can learn more about other ways to get connected and find support. If you need additional support, please contact our Support Navigators at support@tscalliance.org.

FIND A TSC CLINIC
BCUREFUL TRAVEL & LODGING ASSISTANCE
EDUCATIONAL SUPPORT
COMMUNITY SUPPORT NETWORK
ADULT SUPPORT
CONNECT WITH A SUPPORT NAVIGATOR

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(In case you missed it)



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Thank you!

Michael K. Sowell, MD

michael.sowell@louisville.edu







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Case 2: Dravet Syndrome: A Case Study in Evolving Diagnosis & Precision Treatment

A six-year-old male presenting with a clinical picture most compatible with Dravet syndrome — illustrating the diagnostic challenges, treatment course, and ultimate genetic confirmation across nearly two decades of care.

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Diagnostic Workup & Early Seizure Burden



INITIAL PRESENTATION

Seizure Semiology

Multiple seizure types documented: **generalized tonic-clonic**, **focal**, and **myoclonic**—all provoked or worsened by illness, fever, and excitement, consistent with the Dravet phenotype.

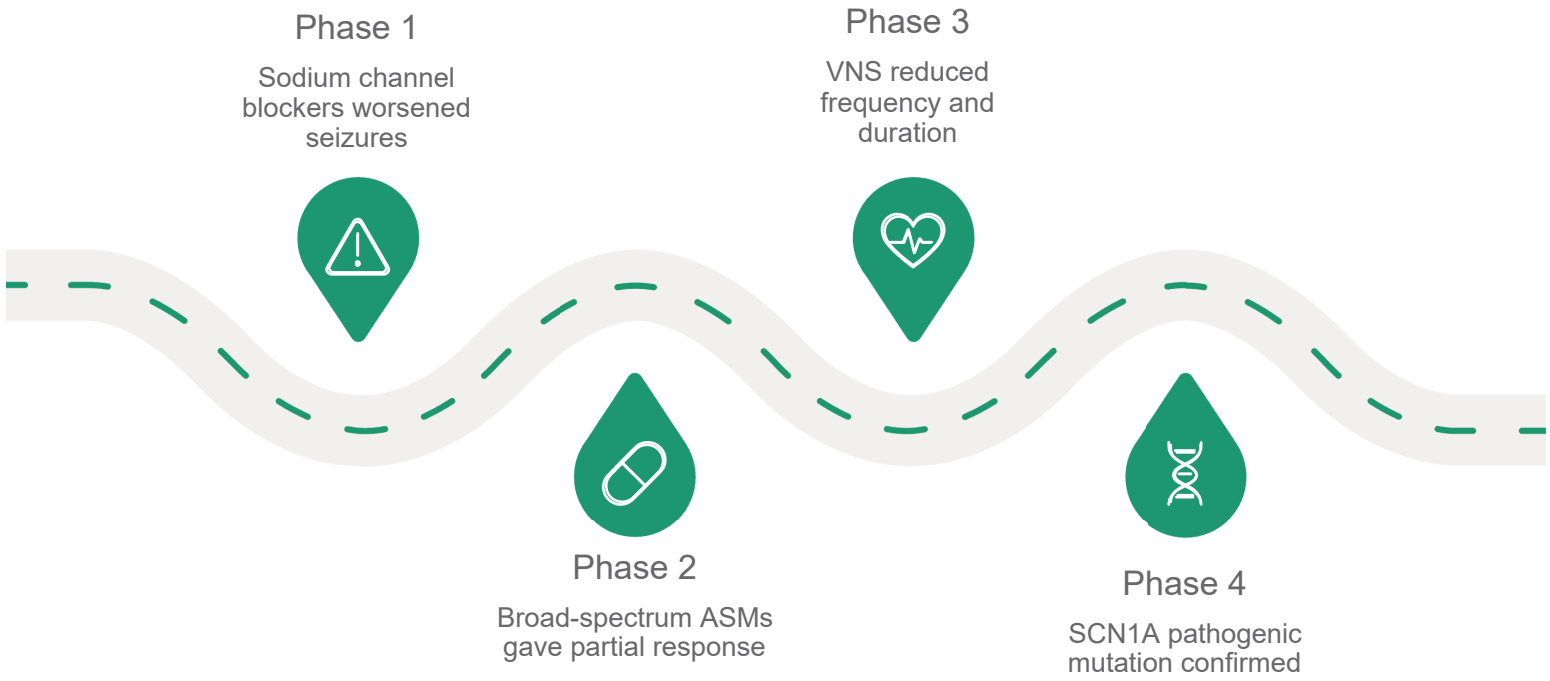
Negative Workup

- **SCN1A genetic testing** negative at age 6; repeat panel at age 10 also negative for known pathogenic mutations
- **Brain MRI** normal
- **Inborn errors of metabolism** normal

A negative SCN1A result does not exclude Dravet syndrome. Clinical diagnosis should guide management when the phenotype is compelling.

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Treatment Trajectory & Precision Medicine Outcome



This case underscores that **sodium channel blockers are contraindicated** in Dravet syndrome and may paradoxically worsen seizures. VNS provided meaningful adjunctive benefit, while genetic re-testing with expanded sequencing ultimately enabled a precision pharmacology approach — highlighting the critical importance of repeat genetic evaluation as technologies evolve.

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Case 3:
19 yoM with GRIN2B

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19yoM with GRIN2B

- Tonic seizures
- History of generalized tonic clonic seizures
- Tonic seizures worsen with “excitement.”
- Anything “exciting” – perseverates on it and can’t sleep.
- Family wants to help him have fewer seizures.

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What has he already tried?

- 9 different anti - seizure medications
- Vagus nerve stimulator
- Prior high dose diazepam for ESES
- Gets Applied Behavior Analysis (ABA) therapy twice weekly

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Excitement could have underlying anxiety

- Should we treat the anxiety? How?
- Meds he's already on:
  - Cannabidiol (Epidiolex)
  - Clobazam
  - Felbamate
  - Lamotrigine
  - Valproic Acid

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Anti-Anxiety drug choice

SSRI- Sertraline

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Neuropsych Testing

2 years later, neuropsychologist reaches out concerned for ADHD.

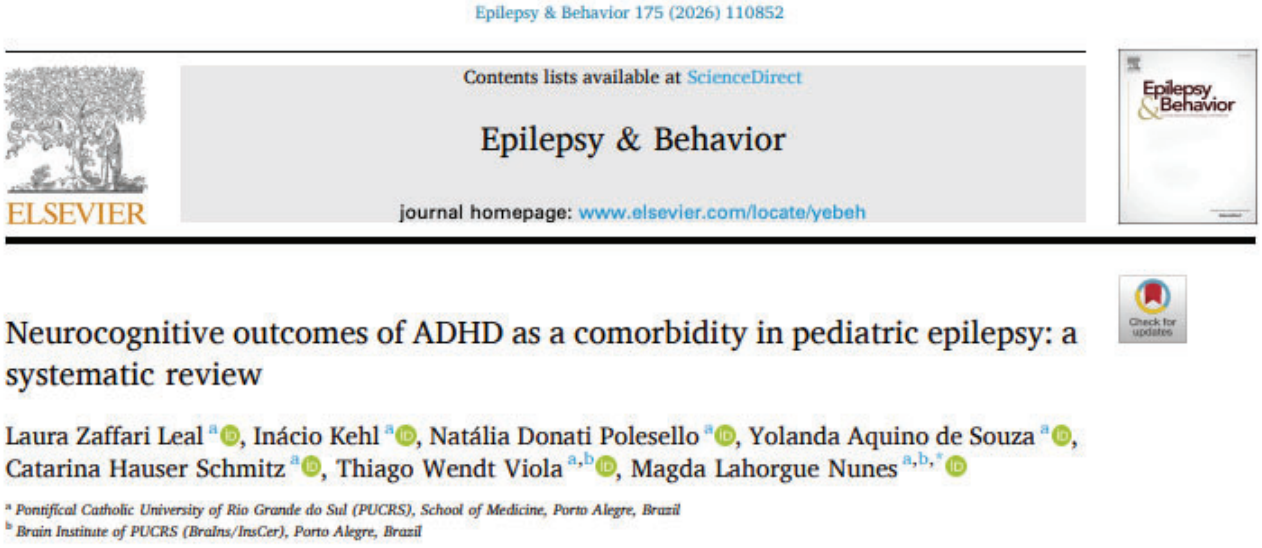
What to try next?

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Discussion about ADHD treatment in Epilepsy

ADHD is common – 8% of general population

- Between 22-30% of children with epilepsy have ADHD.



Epilepsy & Behavior 175 (2026) 110852

Contents lists available at ScienceDirect

Epilepsy & Behavior

journal homepage: www.elsevier.com/locate/yebeh

Neurocognitive outcomes of ADHD as a comorbidity in pediatric epilepsy: a systematic review

Laura Zaffari Leal [a], Inácio Kehl [a], Natália Donati Polesello [a], Yolanda Aquino de Souza [a], Catarina Hauser Schmitz [a], Thiago Wendt Viola [a,b], Magda Lahorgue Nunes [a,b,*]

[a] Pontifical Catholic University of Rio Grande do Sul (PUCRS), School of Medicine, Porto Alegre, Brazil
[b] Brain Institute of PUCRS (BraIns/InsCer), Porto Alegre, Brazil

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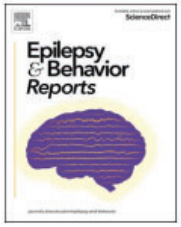
Treatment: Medication + Therapy

Epilepsy & Behavior Reports 29 (2025) 100739

Contents lists available at ScienceDirect

Epilepsy & Behavior Reports

journal homepage: www.elsevier.com/locate/ebcr



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ADHD in Adults with Epilepsy: A Guide for Neurologists☆

Luciana Giambarberi [a,b,*], Halley B. Alexander [a], Heidi M. Munger Clary [a]

[a] Department of Neurology, Wake Forest University School of Medicine, Winston-Salem, NC 27157, USA
[b] Department of Psychiatry, Wake Forest University School of Medicine, Winston-Salem, NC 27103, USA

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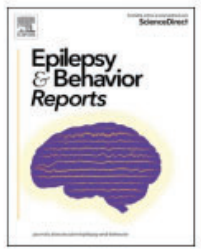
Stimulants are OK in epilepsy

Epilepsy & Behavior Reports 27 (2024) 100679

Contents lists available at ScienceDirect

Epilepsy & Behavior Reports

journal homepage: www.elsevier.com/locate/ebcr



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Do psychotropic drugs cause seizures?

Margaret Gopaul [a,b], Hamada Altalib [a,b,*]

[a] Yale Comprehensive Epilepsy Center, Dept. of Neurology, Yale University School of Medicine New Haven, CT, USA
[b] Veteran Administration (VA) Epilepsy Center of Excellence in West Haven, CT, USA

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You can find all of this information on our website.

Survey- Please Take a Moment

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