

ILAE Academy and EpiCARE

Bibliography to case study: Gabriel (2 y/o) / Tuberous Sclerosis Complex

- Arzimanoglou et al Pediatric Epilepsy Surgery. 2016, John Libbey Eurotext
- Baron Y, Barkovich AJ (1999) MR imaging of tuberous sclerosis in neonates and young infants. *American Journal of NeuroRadiology*. May;20(5):907-16. PMID: 10369365
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7056154/pdf/i0195-6108-020-05-0907.pdf>
- Brodie MJ, Sills GJ. (2011) Combining antiepileptic drugs--rational polytherapy? *Seizure*. Jun;20(5):369-75. doi: 10.1016/j.seizure.2011.01.004.
- Chu-Shore CJ, Major P, Camposano S, Muzykewicz D, Thiele EA. (2010) The natural history of epilepsy in tuberous sclerosis complex. *Epilepsia*. Jul;51(7):1236-41. doi: 10.1111/j.1528-1167.2009.02474.x.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3065368/pdf/nihms277112.pdf>
- Curatolo P, Bombardieri R, Jozwiak S (2008) Tuberous sclerosis. *Lancet*. Aug 23;372(9639):657-68. doi: 10.1016/S0140-6736(08)61279-9.
- Curatolo P (2015) Mechanistic target of rapamycin (mTOR) in tuberous sclerosis complex-associated epilepsy. *Pediatric Neurology*. Mar;52(3):281-9. doi: 10.1016/j.pediatrneurol.2014.10.028.
- Curatolo P, Nabbout R, Lagae L, Aronica E, Ferreira JC, Feucht M, Hertzberg C, Jansen AC, Jansen F, Kotulska K, Moavero R, O'Callaghan F, Papavasiliou A, Tzadok M, Jóźwiak S. (2018) Management of epilepsy associated with tuberous sclerosis complex: Updated clinical recommendations. *European Journal of Pediatric Neurology*. Sep;22(5):738-748. doi: 10.1016/j.ejpn.2018.05.006.
- Cusmai R, Chiron C, Curatolo P, Dulac O, Tran-Dinh S. (1990) Topographic comparative study of magnetic resonance imaging and electroencephalography in 34 children with tuberous sclerosis. *Epilepsia*. Nov-Dec;31(6):747-55. doi: 10.1111/j.1528-1157.1990.tb05516.x.
- Davies M, Saxena A, Kingswood JC (2017) Management of everolimus-associated adverse events in patients with tuberous sclerosis complex: a practical guide. *Orphanet Journal of Rare Diseases*. Feb 15;12(1):35. doi: 10.1186/s13023-017-0581-9.
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5311836/pdf/13023_2017_Article_581.pdf
- Davis PE, Filip-Dhima R, Sideridis G, Peters JM, Au KS, Northrup H, Bebin EM, Wu JY, Krueger D, Sahin M; Tuberous Sclerosis Complex Autism Center of Excellence Research Network. (2017) Presentation and Diagnosis of Tuberous Sclerosis Complex in Infants. *Pediatrics*. Dec;140(6):e20164040. doi: 10.1542/peds.2016-4040.
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5703775/pdf/PEDS_20164040.pdf
- De Ridder J, Lavanga M, Verhelle B, Vervisch J, Lemmens K, Kotulska K, Moavero R, Curatolo P, Weschke B, Riney K, Feucht M, Krsek P, Nabbout R, Jansen AC, Wojdan K, Domanska-Pakieła D, Kaczorowska-Frontczak M, Hertzberg C, Ferrier CH, Samueli S, Benova B, Aronica E, Kwiatkowski DJ, Jansen FE, Jóźwiak S, Van Huffel S, Lagae L. (2020) Prediction of Neurodevelopment in Infants With Tuberous Sclerosis Complex Using Early EEG Characteristics. *Frontiers Neurology*. Oct 16;11:582891. doi: 10.3389/fneur.2020.582891 <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7596378/pdf/fneur-11-582891.pdf>
- De Vries PJ, Belousova E, Benedik MP, Carter T, Cottin V, Curatolo P, D'Amato L, Beure d'Augères G, Ferreira JC, Feucht M, Fladrowski C, Hertzberg C, Jozwiak S, Lawson JA, Macaya A, Marques R, Nabbout R, O'Callaghan F, Qin J, Sander V, Sauter M, Shah S, Takahashi Y, Touraine R, Youroukos S, Zonnenberg B, Kingswood JC, Jansen AC; TOSCA Consortium and TOSCA Investigators. (2020) Natural clusters of tuberous sclerosis complex (TSC)-associated neuropsychiatric disorders (TAND): new findings from the TOSCA TAND research project. *Journal of Neurodevelopmental disorders*. Sep 1;12(1):24. doi: 10.1186/s11689-020-09327-0. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7465404/>

- Ebrahimi-Fakhari D, Agricola KD, Tudor C, Krueger D, Franz DN. (2020) Cannabidiol Elevates Mechanistic Target of Rapamycin Inhibitor Levels in Patients With Tuberous Sclerosis Complex. *Pediatric Neurology*. Apr;105:59-61. doi: 10.1016/j.pediatrneurol.2019.11.017.
- Fallah A, Rodgers SD, Weil AG, Vadera S, Mansouri A, Connolly MB, Major P, Ma T, Devinsky O, Weiner HL, Gonzalez-Martinez JA, Bingaman WE, Najm I, Gupta A, Ragheb J, Bhatia S, Steinbok P, Witiw CD, Widjaja E, Snead OC, Rutka JT. (2015) Resective Epilepsy Surgery for Tuberous Sclerosis in Children: Determining Predictors of Seizure Outcomes in a Multicenter Retrospective Cohort Study. *Neurosurgery*. Oct;77(4):517-24; discussion 524. doi: 10.1227/NEU.0000000000000875. <https://watermark.silverchair.com/neurosurgery-77.4.517.pdf>
- French JA, Lawson JA, Yapici Z, Ikeda H, Polster T, Nabbout R, Curatolo P, de Vries PJ, Dlugos DJ, Berkowitz N, Voi M, Peyrard S, Pelov D, Franz DN. (2016) Adjunctive everolimus therapy for treatment-resistant focal-onset seizures associated with tuberous sclerosis (EXIST-3): a phase 3, randomised, double-blind, placebo-controlled study. *Lancet*. Oct 29;388(10056):2153-2163. doi: 10.1016/S0140-6736(16)31419-2. <https://www.thelancet.com/action/showPdf?pii=S0140-6736%2816%2931419-2>
- Hess EJ, Moody KA, Geffrey AL, Pollack SF, Skirvin LA, Bruno PL, Paolini JL, Thiele EA (2016) Cannabidiol as a new treatment for drug-resistant epilepsy in tuberous sclerosis complex. *Epilepsia*. Oct;57(10):1617-1624. doi: 10.1111/epi.13499. <https://onlinelibrary.wiley.com/doi/epdf/10.1111/epi.13499>
- Jansen FE, Vincken KL, Algra A, Anbeek P, Braams O, Nellist M, Zonnenberg BA, Jennekens-Schinkel A, van den Ouweland A, Halley D, van Huffelen AC, van Nieuwenhuizen O. (2008) Cognitive impairment in tuberous sclerosis complex is a multifactorial condition. *Neurology*. Mar 18;70(12):916-23. doi: 10.1212/01.wnl.0000280579.04974.c0.
- Jayakar P, Gaillard WD, Tripathi M, Libenson MH, Mathern GW, Cross JH; Task Force for Paediatric Epilepsy Surgery, Commission for Paediatrics, and the Diagnostic Commission of the International League Against Epilepsy (2014) Diagnostic test utilization in evaluation for resective epilepsy surgery in children. *Epilepsia*. Apr;55(4):507-18. doi: 10.1111/epi.12544. <https://onlinelibrary.wiley.com/doi/epdf/10.1111/epi.12544>
- Kotulska K, Kwiatkowski DJ, Curatolo P, Weschke B, Riney K, Jansen F, Feucht M, Krsek P, Nabbout R, Jansen AC, Wojdan K, Sijko K, Głowacka-Walas J, Borkowska J, Sadowski K, Domańska-Pakieła D, Moavero R, Hertzberg C, Hulshof H, Scholl T, Benova B, Aronica E, de Ridder J, Lagae L, Jóźwiak S; EPISTOP Investigators. (2021) Prevention of Epilepsy in Infants with Tuberous Sclerosis Complex in the EPISTOP Trial. *Annals of Neurology*. Feb;89(2):304-314. doi: 10.1002/ana.25956. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7898885/pdf/ANA-89-304.pdf>
- Kwan P, Arzimanoglou A, Berg AT, Martin JB, Hauser WA, Mathern G, Moshé SL, Perucca E, Wiebe S, French J (2010) Definition of drug resistant epilepsy: consensus proposal by the ad hoc Task Force of the ILAE commission on therapeutic strategies. *Epilepsia*. Jun;51(6):1069-77. Doi 10.1111/j.15861167.2009.02397.x. <https://onlinelibrary.wiley.com/doi/epdf/10.1111/j.1528-1167.2009.02397.x>
- Moavero R, Benvenuto A, Emberti Gialloreti L, Siracusano M, Kotulska K, Weschke B, Riney K, Jansen FE, Feucht M, Krsek P, Nabbout R, Jansen AC, Wojdan K, Borkowska J, Sadowski K, Hertzberg C, Hulshof H, Samuelli S, Benova B, Aronica E, Kwiatkowski DJ, Lagae L, Jozwiak S, Curatolo P. (2019) Early Clinical Predictors of Autism Spectrum Disorder in Infants with Tuberous Sclerosis Complex: Results from the EPISTOP Study. *Journal of Clinical Medicine*. Jun 3;8(6):788. doi: 10.3390/jcm8060788. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6617179/pdf/jcm-08-00788.pdf>
- Mühlebner A, Bongaarts A, Sarnat HB, Scholl T, Aronica E. (2019) New insights into a spectrum of developmental malformations related to mTOR dysregulations: challenges and perspectives. *Journal of Anatomy*. Sep;235(3):521-542. doi: 10.1111/joa.12956. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6704243/pdf/JOA-235-521.pdf>

- Nabbout R, Belousova E, Benedik MP, Carter T, Cottin V, Curatolo P, Dahlin M, Amato LD, Beure d'Augères G, de Vries PJ, Ferreira JC, Feucht M, Fladrowski C, Hertzberg C, Jozwiak S, Lawson JA, Macaya A, Marques R, O'Callaghan F, Qin J, Sander V, Sauter M, Shah S, Takahashi Y, Touraine R, Youroukos S, Zonnenberg B, Jansen A, Kingswood JC, TOSCA Consortium and TOSCA Investigators (2018) Epilepsy in tuberous sclerosis complex: Findings from the TOSCA Study. *Epilepsia Open*. Dec 21;4(1):73-84. doi: 10.1002/epi4.12286.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6398114/pdf/EPI4-4-73.pdf>
- Napolioni V, Moavero R, Curatolo P. (2009) Recent advances in neurobiology of Tuberous Sclerosis Complex. *Brain Development*. Feb;31(2):104-13. doi: 10.1016/j.braindev.2008.09.013.
- Neal A, Ostrowsky-Coste K, Jung J, Lagarde S, Maillard L, Kahane P, Touraine R, Catenoix H, Montavont A, Isnard J, Arzimanoglou A, Bartolomei F, Guenot M, Rheims S. (2019) Epileptogenicity in tuberous sclerosis complex: A stereoelectroencephalographic study. *Epilepsia*. Jan;61(1):81-95. doi: 10.1111/epi.16410. Epub 2019 Dec 20.
- Northrup H, Aronow ME, Bebin EM, et al. (2021). Updated International Tuberous Sclerosis Complex Diagnostic Criteria and Surveillance and Management Recommendations. *Pediatr Neurol*. 2021;123:50-66. doi: 10.1016/j.pediatrneurol.2021.07.011
- Northrup H, Krueger DA; International Tuberous Sclerosis Complex Consensus Group. (2013) Tuberous sclerosis complex diagnostic criteria update: recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. *Pediatric Neurology*. Oct;49(4):243-54. doi: 10.1016/j.pediatrneurol.2013.08.001.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4080684/pdf/nihms587050.pdf>
- Ostrowsky-Coste K, Neal A, Guenot M, Ryvlin P, Bouvard S, Bourdillon P, Jung J, Catenoix H, Montavont A, Isnard J, Arzimanoglou A, Rheims S. (2019) Resective surgery in tuberous Sclerosis complex, from Penfield to 2018: A critical review. *Revue Neurologique*. Mar;175(3):163-182. doi: 10.1016/j.neurol.2018.11.002.
- Rosenberg EC, Tsien RW, Whalley BJ, Devinsky O (2015) Cannabinoids and Epilepsy. *Neurotherapeutics*. Oct;12(4):747-68. doi: 10.1007/s13311-015-0375-5.
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4604191/pdf/13311_2015_Article_375.pdf
- Russo C, Nastro A, Cicala D, De Liso M, Covelli EM, Cinalli G. (2020) Neuroimaging in tuberous sclerosis complex. *Child Nervous System*. Oct;36(10):2497-2509. doi: 10.1007/s00381-020-04705-4.
- Słowińska M, Józwiak S, Peron A, Borkowska J, Chmielewski D, Sadowski K, Jurkiewicz E, Vignoli A, La Briola F, Canevini MP, Kotulska-Józwiak K. (2018) Early diagnosis of tuberous sclerosis complex: a race against time. How to make the diagnosis before seizures? *Orphanet Journal of Rare Diseases*. Jan 29;13(1):25. doi: 10.1186/s13023-018-0764-z.
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5789613/pdf/13023_2018_Article_764.pdf
- Thiele EA, Bebin EM, Bhathal H, Jansen FE, Kotulska K, Lawson JA, O'Callaghan FJ, Wong M, Sahebkar F, Checketts D, Knappertz V; GWPCARE6 Study Group. (2021) Add-on Cannabidiol Treatment for Drug-Resistant Seizures in Tuberous Sclerosis Complex: A Placebo-Controlled Randomized Clinical Trial. *JAMA Neurology*. Mar 1;78(3):285-292. doi: 10.1001/jamaneurol.2020.4607.
<https://jamanetwork.com/journals/jamaneurology/fullarticle/2774314>
- Tyburczy ME, Dies KA, Glass J, Camposano S, Chekaluk Y, Thorner AR, Lin L, Krueger D, Franz DN, Thiele EA, Sahin M, Kwiatkowski DJ (2015) Mosaic and Intronic Mutations in TSC1/TSC2 Explain the Majority of TSC Patients with No Mutation Identified by Conventional Testing. *PLoS genetics*. Nov 5;11(11):e1005637. doi: 10.1371/journal.pgen.1005637.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4634999/pdf/pgen.1005637.pdf>

- Von Ranke FM, Faria IM, Zanetti G, Hochhegger B, Souza AS Jr, Marchiori E. (2017) Imaging of tuberous sclerosis complex: a pictorial review. *Radiol Bras*. Jan-Feb;50(1):48-54. doi: 10.1590/0100-3984.2016.0020. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5347503/pdf/rb-50-01-0048.pdf>
- Wu JY, Goyal M, Peters JM, Krueger D, Sahin M, Northrup H, Au KS, O'Kelley S, Williams M, Pearson DA, Hanson E, Byars AW, Krefting J, Beasley M, Cutter G, Limdi N, Bebin EM. (2019) Scalp EEG spikes predict impending epilepsy in TSC infants: A longitudinal observational study. *Epilepsia*. Dec;60(12):2428-2436. doi: 10.1111/epi.16379. Epub 2019 Nov 5. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6910957/pdf/EPI-60-2428.pdf>

Further reading:

[ILAE Website Diagnostic manual](#)

<https://www.epilepsydiagnosis.org/>