

ILAE Academy and EpiCARE

Bibliography to case study: Aurora (7 m/o) / Spasms

- Barba C, Mai R, Grisotto L, Gozzo F, Pellacani S, Tassi L, Francione S, Giordano F, Cardinale F, Guerrini R. Unilobar surgery for symptomatic epileptic spasms. *Ann Clin Transl Neurol*. 2016 Nov 19;4(1):36-45. doi: 10.1002/acn3.373. PMID: 28078313; PMCID: PMC5221449.
- Caputo D, Trivisano M, Vigeveno F, Fusco L. CHD2-epilepsy: Polygraphic documentation of self-induced seizures due to fixation-off sensitivity. *Seizure*. 2018 Apr;57:8-10.
- Chen J, Zhang J, Liu A, Zhang L, Li H, Zeng Q, Yang Z, Yang X, Wu X, Zhang Y. CHD2-related epilepsy: novel mutations and new phenotypes. *Dev Med Child Neurol*. 2020 May;62(5):647-653.
- Datta AN, Bahi-Buisson N, Bienvenu T, Buerki SE, Gardiner F, Cross JH, Heron B, Kaminska A, Korff CM, Lepine A, Lesca G, McTague A, Mefford HC, Mignot C, Milh M, Piton A, Pressler RM, Ruf S, Sadleir LG, de Saint Martin A, Van Gassen K, Verbeek NE, Ville D, Villeneuve N, Zacher P, Scheffer IE, Lemke JR. The phenotypic spectrum of X-linked, infantile onset ALG13-related developmental and epileptic encephalopathy. *Epilepsia*. 2021 Feb;62(2):325-334. doi: 10.1111/epi.16761. Epub 2021 Jan 7. PMID: 33410528; PMCID: PMC7898319.
- de Palma L, Boniver C, Cassina M, Toldo I, Nosadini M, Clementi M, Sartori S. Eating-induced epileptic spasms in a boy with MECP2 duplication syndrome: insights into pathogenesis of genetic epilepsies. *Epileptic Disord*. 2012 Dec;14(4):414-7. doi: 10.1684/epd.2012.0546. PMID: 23248047.
- Fusco, L., Serino, D., Santarone, M.E., 2020. Three different scenarios for epileptic spasms. *Epilepsy Behav*. 113, 107531. <https://doi.org/10.1016/j.yebeh.2020.107531>.
- Gaillard WD, Weinstein S, Conry J, Pearl PL, Fazilat S, Fazilat S, Vezina LG, Reeves-Tyer P, Theodore WH. Prognosis of children with partial epilepsy: MRI and serial 18FDG-PET. *Neurology*. 2007 Feb 27;68(9):655-9. doi: 10.1212/01.wnl.0000255942.25101.8d. PMID: 17325271.
- Govil-Dalela T, Kumar A, Behen ME, Chugani HT, Juhász C. Evolution of lobar abnormalities of cerebral glucose metabolism in 41 children with drug-resistant epilepsy. *Epilepsia*. 2018 Jul;59(7):1307-1315. doi: 10.1111/epi.14404. Epub 2018 May 22. PMID: 29786852; PMCID: PMC6031462.
- Grinspan ZM, Knupp KG, Patel AD, Yozawitz EG, Wusthoff CJ, Wirrell E, Valencia I, Singhal NS, Nordli DR, Mytinger JR, Mitchell W, Keator CG, Loddenkemper T, Hussain SA, Harini C, Gaillard WD, Fernandez IS, Coryell J, Chu CJ, Berg AT, Shellhaas RA. Comparative Effectiveness of Initial Treatment for Infantile Spasms in a Contemporary US Cohort. *Neurology*. 2021 Jul 15;97(12):e1217-28. doi: 10.1212/WNL.0000000000012511
- Howell KB, Freeman JL, Mackay MT, et al. The severe epilepsy syndromes of infancy: A population-based study. *Epilepsia*. 2021 Feb;62(2):358-370. doi: 10.1111/epi.16810.
- Jonas R, Asarnow RF, LoPresti C, Yudovin S, Koh S, Wu JY, Sankar R, Shields WD, Vinters HV, Mathern GW. Surgery for symptomatic infant-onset epileptic encephalopathy with and without infantile spasms. *Neurology*. 2005 Feb 22;64(4):746-50. doi: 10.1212/01.WNL.0000151970.29205.70. PMID: 15728309.
- 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-Wałas 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. Prevention of Epilepsy in Infants with Tuberous Sclerosis Complex in the EPISTOP Trial. *Ann Neurol*. 2021 Feb;89(2):304-314. doi: 10.1002/ana.25956. Epub 2020 Nov 27. PMID: 33180985; PMCID: PMC7898885
- Lamberink HJ, Otte WM, Blümcke I, Braun KPJ; European Epilepsy Brain Bank writing group; study group; European Reference Network EpiCARE. Seizure outcome and use of antiepileptic drugs after epilepsy surgery according to histopathological diagnosis: a retrospective multicentre cohort study. *Lancet Neurol*. 2020 Sep;19(9):748-757. doi: 10.1016/S1474-4422(20)30220-9. PMID: 32822635
- Mei D, Cetica V, Marini C, Guerrini R. Dravet syndrome as part of the clinical and genetic spectrum of sodium channel epilepsies and encephalopathies. *Epilepsia*. 2019 Dec;60 Suppl 3:S2-S7.

- Muir, A.M., Myers, C.T., Nguyen, N.T., et al 2019. Genetic heterogeneity in infantile spasms. *Epilepsy Res.* 156, 106181. <https://doi.org/10.1016/j.eplepsyres.2019.106181>
- National Clinical Guideline Centre, National Institute for Health and Clinical Excellence. The epilepsies. The diagnosis and management of the epilepsies in adults and children in primary and secondary care. Pharmacological update of clinical guideline 20. Final. Methods, evidence and recommendations. London: Royal College of Physicians; 2012.
- O'Callaghan FJ, Edwards SW, Alber FD, Hancock E, Johnson AL, Kennedy CR, Likeman M, Lux AL, Mackay M, Mallick AA, Newton RW, Nolan M, Pressler R, Rating D, Schmitt B, Verity CM, Osborne JP; participating investigators. Safety and effectiveness of hormonal treatment versus hormonal treatment with vigabatrin for infantile spasms (ICISS): a randomised, multicentre, open-label trial. *Lancet Neurol.* 2017 Jan;16(1):33-42. doi: 10.1016/S1474-4422(16)30294-0. Epub 2016 Nov 10. PMID: 27838190.
- Salar S, Moshé SL, Galanopoulou AS. Metabolic etiologies in West syndrome. *Epilepsia Open.* 2018 Mar 14;3(2):134-166. doi: 10.1002/epi4.12102. PMID: 29881795; PMCID: PMC5983207.
- SoRelle, J.A., Thodeson, D.M., Arnold, S., Gotway, G., Park, J.Y., 2019. Clinical Utility of Reinterpreting Previously Reported Genomic Epilepsy Test Results for Pediatric Patients. *JAMA Pediatr.* 173, e182302.
- Specchio N, Pietrafusa N, Ferretti A, De Palma L, Santarone ME, Pepi C, Trivisano M, Vigeveno F, Curatolo P. Treatment of infantile spasms: why do we know so little? *Expert Rev Neurother.* 2020 Jun;20(6):551-566. doi: 10.1080/14737175.2020.1759423. Epub 2020 Jun 19. PMID: 32316776.