

## ILAE Academy and EpiCARE

### Bibliography to case study: Jules (Dravet Syndrome)

#### General

Arzimanoglou A, O'Hare A, Johnston VM, Ouvrier R (2018). *London: Mc Keith Press: Aicardi's disease of the nervous system in children, (4<sup>th</sup> Edition)*, 847-996

Bureau M, Genton P, Dravet C, Delgado-Escueta AV, Tassinari CA, Thomas P, Wolf P (2013). *John Libbey Eurotext: Epileptic syndromes in infancy, childhood and adolescence, (5<sup>th</sup> Edition)*, 125-156

Dravet C, Guerrini R (2011). *Dravet Syndrome. John Libbey Eurotext: Topics in epilepsy*, 12-32.

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.

<https://www.nice.org.uk/guidance/cg137/resources/epilepsies-diagnosis-and-management-pdf-35109515407813>

#### Dravet – diagnosis

Arsalan A, Sidra S, Ur Vish KP, Kogulavadanan A, Preeti M (2019). Dravet syndrome: an Overview. *Cureus*, Jun 26;11(6):e5006. doi: 10.7759/cureus.5006

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6713249/pdf/cureus-0011-0000005006.pdf>

Arzimanoglou A (2009). Dravet syndrome: from electroclinical characteristics to molecular biology. *Epilepsia*, 50(Suppl 8):3-9. doi: 10.1111/j.1528-1167.2009.02228.x

<https://onlinelibrary.wiley.com/doi/epdf/10.1111/j.1528-1167.2009.02228.x>

Bureau M, Dalla Bernardina B (2011). Electroencephalographic characteristics of Dravet syndrome. *Epilepsia*, 52(Suppl 2):13-23. doi: 10.1111/j.1528-1167.2011.02996.x

<https://onlinelibrary.wiley.com/doi/epdf/10.1111/j.1528-1167.2011.02996.x>

Chemaly N, Losito E, Pinard JM, Gautier A, Villeneuve N, Arbues AS, An I, Desguerre I, Dulac O, Chiron C, Kaminska A, Nabbout R (2018). Early and long-term electroclinical features of patients with epilepsy and PCDH19 mutation. *Epileptic Disorders*, 20(6):457-67. doi: 10.1684/epd.2018.1009

[https://www.jle.com/download/epd-313315-40950-early\\_and\\_long\\_term\\_electroclinical\\_features\\_of\\_patients\\_with\\_epilepsy\\_and\\_pcdh19\\_mutation-a.pdf](https://www.jle.com/download/epd-313315-40950-early_and_long_term_electroclinical_features_of_patients_with_epilepsy_and_pcdh19_mutation-a.pdf)

Darra F, Battaglia D, Dravet C, Patrini M, Offredi F, Chieffo D, Piazza E, Fontana E, Olivieri G, Turrini I, Dalla Bernardina B, Granata T, Ragona F (2019). Dravet syndrome: Early electroclinical findings and long-term outcome in adolescents and adults. *Epilepsia*, Dec;60(Suppl 3):S49-S58. doi: 10.1111/epi.16297

<https://onlinelibrary.wiley.com/doi/epdf/10.1111/epi.16297>

Dravet C (2011). The core Dravet syndrome phenotype. *Epilepsia* Apr;52(Suppl 2):3-9. doi: 10.1111/j.1528-1167.2011.02994.x

<https://onlinelibrary.wiley.com/doi/epdf/10.1111/j.1528-1167.2011.02994.x>

Frucht MM, Quigg M, Schwaner C, Fountain NB (2000). Distribution of seizure precipitants among epilepsy syndromes. *Epilepsia*, 41(12):1534-1539. doi: 10.1111/j.1499-1654.2000.001534.x

<https://onlinelibrary.wiley.com/doi/epdf/10.1111/j.1499-1654.2000.001534.x>

Kolc KL, Sadleir LG, Scheffer IE, Ivancevic A, Roberts R, Pham DH, Gecz J (2019). A systematic review and meta-analysis of 271 PCDH19-variant individuals identifies psychiatric comorbidities, and association of seizure onset and disease severity. *Molecular Psychiatry*, 24:241-251. doi:

10.1038/s41380-018-0066-9

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7198503/>

Lamperti C, Zeviani M (2016). Myoclonus epilepsy in mitochondrial disorders. *Epileptic disorders*, 18(Suppl2):S94-S102. doi: 10.1684/epd.2016.0846

Marini C, Scheffer IE, Nabbout R, Suls A, De Jonghe P, Zara Z, Guerrini R (2011). The genetics of Dravet syndrome. *Epilepsia*, Apr;52 (Suppl 2):24-9. doi: 10.1111/j.1528-1167.2011.02997.x.

<https://onlinelibrary.wiley.com/doi/epdf/10.1111/j.1528-1167.2011.02997.x>

Ouss L, Leunen D, Laschet J, Chemaly N, Barcia G, Losito EM, Aouidad A, Barrault Z, Desguerre I, Breuillard D, Nabbout R (2019). Autism spectrum disorder and cognitive profile in children with Dravet syndrome : Delineation of a specific phenotype. *Epilepsia Open*, 4:40-53. doi: 10.1002/epi4.12281

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6398110/pdf/EPI4-4-40.pdf>

Pardo CA, Nabbout R, Galanopoulou AS (2014). Mechanisms of epileptogenesis in pediatric epileptic syndromes: Rasmussen encephalitis, Infantile spasms, and Febrile Infection related epilepsy syndrome (FIRES). *Neurotherapeutics*, 11:297-310. doi: 10.1007/s13311-014-0265-2

Scheffer IE, Nabbout R (2019). SCN1A-related phenotypes: Epilepsy and beyond. *Epilepsia*, Dec;60 (Suppl 3):S17-S24. doi: 10.1111/epi.16386

<https://onlinelibrary.wiley.com/doi/epdf/10.1111/epi.16386>

Steel D, Symonds JD, Zuberi SM, Brunklaus A (2017). Dravet syndrome and its mimics: Beyond SCN1A. *Epilepsia*, 58(11):1807-1816. doi: 10.1111/epi.13889

<https://onlinelibrary.wiley.com/doi/epdf/10.1111/epi.13889>

Van Campen JS, Jansen FE, Steinbusch LC, Joels M, Braun KPJ (2012). Stress sensitivity of childhood epilepsy is related to experience negative life events. *Epilepsia*, 59(9):1554-1562. doi: 10.1111/j.1528-1167.2012.03566.x

### **Dravet counselling & SUDEP**

Genton P, Velizarova R, Dravet C (2011). Dravet syndrome: the long term outcome. *Epilepsia*, 52(Suppl 2):44-49. doi: 10.1111/j.1528-1167.2011.03001

<https://onlinelibrary.wiley.com/doi/epdf/10.1111/j.1528-1167.2011.03001.x>

Guerrini R, Falchi M (2011). Dravet syndrome and SCN1A gene mutation related epilepsies: cognitive impairment and its determinants. *Developmental medicine & Child Neurology*, 53(Suppl, 2):11-15. doi: 10.1111/j.1469-8749.2011.03966.x

<https://onlinelibrary.wiley.com/doi/epdf/10.1111/j.1469-8749.2011.03966.x>

Jansson JS, Hallböök T, Reilly C (2020). Intellectual functioning and behavior in Dravet syndrome: A systematic review. *Epilepsy Behaviour*, Apr 22;108:107079. doi: 10.1016/j.yebeh.2020.107079.

Maguire MJ, Jackson CF, Marson AG, Nevitt SJ (2020). Treatments for the prevention of Sudden Unexpected Death in Epilepsy (SUDEP) (Review). *Cochrane database of Systematic Review*, issue 4 art. No DC011792. doi: 10.1002/14651858.CD011792.pub3.

Nabbout R, Chemaly N, Chipaux M, Barcia G, Bouis C, Dubouch C, Leunen D, Jambaqué I, Dulac O, Dellatolas G, Chiron C (2013). Encephalopathy in Children With Dravet Syndrome Is Not a Pure Consequence of Epilepsy. *Orphanet J Rare Dis.*, Nov 13;8:176. doi: 10.1186/1750-1172-8-176

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4225757/pdf/1750-1172-8-176.pdf>

Ragona F (2011). Cognitive development in children with Dravet syndrome. *Epilepsia*, 52(Suppl 2):39-43. doi: 10.1111/j.1528-1167.2011.03000.x

<https://onlinelibrary.wiley.com/doi/epdf/10.1111/j.1528-1167.2011.03000.x>

Sakauchi M, Oguni H, Kato I, Osawa M, Hirose S, Kaneko S, Takahashi Y, Takayama R, Fujiwara T (2011). Mortality in Dravet syndrome: search for risk factor in Japanese patients. *Epilepsia*, 52(Suppl 2):50-54. doi: 10.1111/j.1528-1167.2011.03002.x

Shmueli S, Sisodiya SM, Gunning WB, Sander JW, Thijs RD (2016). Mortality in Dravet syndrome: a review. *Epilepsy and Behaviour*, 64: 69-74. doi: 10.1016/j.yebeh.2016.09.007  
<https://www.epilepsybehavior.com/action/showPdf?pii=S1525-5050%2816%2930373-0>

### **Treatment interactions – Dravet management**

Arzimanoglou A, Brandl U, Cross JH, Gil-Nagel A, Lagae L, Landmark CJ, Specchio N, Nabbout R, Thiele EA, Gubbay O, The Cannabinoids International Experts Panel (2020). Epilepsy and cannabidiol: a guide to treatment. *Epileptic Disorders*, Feb 1;22(1):1-14. doi: 10.1684/epd.2020.1141.  
[https://www.jle.com/download/epd-316272-44751-epilepsy\\_and\\_cannabidiol\\_a\\_guide\\_to\\_treatment-a.pdf](https://www.jle.com/download/epd-316272-44751-epilepsy_and_cannabidiol_a_guide_to_treatment-a.pdf)

Chiron C, Dulac O (2011). The pharmacological treatment of Dravet syndrome. *Epilepsia*, 2011 Apr 52 (Suppl 2):72-5. doi:10.1111/j.1528-1167.2011.03007.x

Cross JH, Caraballo RH, Nabbout R, Vigeveno F, Guerrini R, Lagae L (2019). Dravet syndrome : treatment options and management of prolonged seizures. *Epilepsia*, 60(S3):S39-S48. doi: 10.1111/epi.16334  
<https://onlinelibrary.wiley.com/doi/epdf/10.1111/epi.16334>

Devinsky O, Cross JH, Laux L, Marsh E, Miller I, Nabbout R, Scheffer IE, Thiele EA, Wright S “Cannabidiol in Dravet Syndrome Study Group” (2017). Trial of Cannabidiol for Drug-Resistant Seizures in the Dravet Syndrome. *New England Journal of Medicine*, May 25;376(21):2011-2020. doi: 10.1056/NEJMoa1611618.  
<https://www.nejm.org/doi/pdf/10.1056/NEJMoa1611618?articleTools=true>

Devinsky O, Verducci C, Thiele EA, Laux LC, Patel AD, Filloux F, Szaflarski JP, Wilfong A, Clark GD, ParkYD, Seltzer LE, Bebin EM, Flamini R, Wechsler RT, Friedman D (2018). Open-label use of highly purified CBD (Epidiolex®) in patients with CDKL5 deficiency disorder and Aicardi, Dup15q, and Doose syndromes. *Epilepsy Behaviour*, Sep;86:131-137. doi: 10.1016/j.yebeh.2018.05.013  
<https://www.epilepsybehavior.com/action/showPdf?pii=S1525-5050%2818%2930191-4>

Fischer JL (2011). The effects of stiripentol on GABA(A) receptors. *Epilepsia*, Apr;52 (Suppl 2):76-78. doi: 10.1111/j.1528-1167.2011.03008.x

Garnett WR (2000). Clinical pharmacology of topiramate: a review. *Epilepsia*, 41(Suppl 1):S61-S65. doi: 10.1111/j.1528.1157.2000.tb02174.x

Lagae L, Brambilla I, Mingorance A, Gibson E, Battersby A (2018). Quality of life and comorbidities associated with Dravet syndrome severity: a multinational cohort survey. *Developmental medicine and Child Neurology*, 60:63-72. doi: 10.1111/dmcn.13591  
<https://onlinelibrary.wiley.com/doi/epdf.10.1111/dmcn.13591>

Patsalos PN, Berry DJ, Bourgeois BFD, CLoyd JC, Glauser TA, Johannessen SI, Leppik IE, Tomson T, Perrucca E (2008). Antiepileptic drugs – Best practice guidelines for therapeutic drug monitoring: A position paper by the subcommission on therapeutic drug monitoring, ILAE Commission on Therapeutic Strategies. *Epilepsia*, 49(7):1239-1276. doi: 10.1111:j.1528-1167.2008.01561.x  
<https://onlinelibrary.wiley.com/doi/epdf/10.1111/j.1528-1167.2008.01561.x>

Perucca E (1996). Pharmacokinetic profile of topiramate in comparison with other new antiepileptic drugs. *Epilepsia*, 37 (Suppl. 2):S8-S13. doi: 10.1111/j.1528-1157.1996.tb06032.x

Rosenberg EC, Tsien RW, Whalley BJ, Devinsky O (2015). Cannabinoids and epilepsy. *Neurotherapeutics*, Oct;12(4):747-68. doi: 10.1007/s.13311-015-0375-5

## **Differential diagnosis**

Darra F, Fiorini A, Zoccante L, Mastella L, Torniero C, Cortese S, Meneghello L, Fontana E, Dalla Bernardina B (2006). Benign Myoclonic Epilepsy in Infancy (BMEI): a longitudinal electroclinical study of 22 cases. *Epilepsia*, 47(Suppl 5):31-5. doi: 10.1111/j.1528-1167.2006.00874.x  
<https://onlinelibrary.wiley.com/doi/epdf/10.1111/j.1528-1167.2006.00874.x>

Rahman S (2012). Mitochondrial disease and epilepsy. *Developmental Medicine and Child Neurology*, May;54(5):397-406. doi: 10.1111/j.1469-8749.2011.04214.x  
<https://onlinelibrary.wiley.com/doi/epdf/10.1111/j.1469-8749.2011.04214.x>

Williams RE, Aberg L, Autti T, Goebel HH, Kohlschütter A, Lönnqvist L (2006). Diagnosis of the neuronal ceroid lipofuscinoses: an update. *Biochimica and Biophysica Acta*, Oct;1762(10):865-72. doi: 10.1016/j.bbadis.2006.07.001  
<https://reader.elsevier.com/reader/sd/pii/S0925443906001281?token=BB4009A602BA754BEA61B63A0A70C707A18DB5CB6F19CBF4BFBD7EA2F76B47CF0DE45A7B952DAF4F5DF4089E467B9D3>