

## ILAE Academy and EpiCARE

### Bibliography to case study: Angela (Autoimmune Encephalitis)

#### REFERENCES (IN ALPHABETIC ORDER)

##### Books

Bureau M, Genton P, Delgado-Escueta A, Dravet C, Guerrini R, Tassinari CA, Thomas P, Wolf P. (2019) *Epileptic Syndromes in Infancy, Childhood and Adolescence*. John Libbey Eurotext.

Guerrini R, Marini C, Barba C. Generalized epilepsies. *Handb Clin Neurol*. 2019;161:3-15.

##### Articles

De Giorgis V, Masnada S, Varesio C, Chiappedi MA, Zanaboni M, Pasca L, Filippini M, Macasaet JA, Valente M, Ferraris C, Tagliabue A, Veggiotti P (2019). Overall cognitive profiles in patients with GLUT1 Deficiency Syndrome. *Brain Behaviour*. Mar;9(3):e01224. doi: 10.1002/brb3.1224  
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6422708/pdf/BRB3-9-e01224.pdf>

De Giorgis V, Varesio C, Baldassari C, Piazza E, Olivotto S, Macasaet J, Balottin U, Veggiotti P (2016). Atypical Manifestations in Glut1 Deficiency Syndrome. *Journal of Child Neurology*. Aug;31(9):1174-80. doi: 10.1177/0883073816650033.

Garone G, Capuano A, Travaglini L, Graziola F, Stregapede F, Zanni G, Vigevano F, Bertini E, Nicita F (2020). Clinical and Genetic Overview of Paroxysmal Movement Disorders and Episodic Ataxias. *International Journal of Molecular Sciences*. May 20;21(10):3603. doi: 10.3390/ijms21103603.  
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7279391/pdf/ijms-21-03603.pdf>

Gavrilovici C, Rho JM (2020). Metabolic epilepsies amenable to ketogenic therapies: Indications, contraindications, and underlying mechanisms. *Journal of Inherited Metabolic Diseases*. Jul 12. doi : 10.1002/jimd.12283

Hafsi W, Badri T (2020). Cockayne Syndrome. Aug 16. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2020 Jan.

Kang HC, Chung DE, Kim DW, Kim HD (2004). Early- and late-onset complications of the ketogenic diet for intractable epilepsy. *Epilepsia*. Sep;45(9):1116-23. doi: 10.1111/j.0013-9580.2004.10004.x.  
<https://onlinelibrary.wiley.com/doi/epdf/10.1111/j.0013-9580.2004.10004.x>

Klepper J, Akman C, Armeno M, Auvin S, Cervenka M, Cross HJ, De Giorgis V, Della Marina A, Engelstad K, Heussinger N, Kossoff EH, Leen WG, Leiendecker B, Monani UR, Oguni H, Neal E, Pascual JM, Pearson TS, Pons R, Scheffer IE, Veggiotti P, Willemsen M, Zuberi SM, De Vivo DC (2020). Glut1 Deficiency Syndrome (Glut1DS): State of the art in 2020 and recommendations of the international Glut1DS study group. *Epilepsia Open*. Aug 13;5(3):354-365. doi: 10.1002/epi4.12414.  
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7469861/pdf/EPI4-5-354.pdf>

Kossoff EH, Veggiotti P, Genton P, Desguerre I (2014). Transition for patients with epilepsy due to metabolic and mitochondrial disorders. *Epilepsia*. Aug;55 Suppl 3:37-40. doi: 10.1111/epi.12709.  
<https://onlinelibrary.wiley.com/doi/epdf/10.1111/epi.12709>

Kossoff EH, Zupec-Kania BA, Amark PE, et al (2009). Optimal clinical management of children receiving the ketogenic diet: recommendations of the International Ketogenic Diet Study Group. *Epilepsia*. Feb;50(2):304-17. doi: 10.1111/j.1528-1167.2008.01765.x. <https://onlinelibrary.wiley.com/doi/epdf/10.1111/j.1528-1167.2008.01765.x>

Leary LD, Wang D, Nordli DR, Engelstad K, De Vivo DC (2003). Seizure characterization and electroencephalographic features in Glut-1 deficiency syndrome. *Epilepsia*. May;44(5):701-7. doi: 10.1046/j.1528-1157.2003.05302.x. <https://onlinelibrary.wiley.com/doi/epdf/10.1046/j.1528-1157.2003.05302.x>

Leen WG, de Wit CJ, Wevers RA, van Engelen BG, Kamsteeg EJ, Klepper J, Verbeek MM, Willemsen MA (2013). Child neurology: differential diagnosis of a low CSF glucose in children and young adults. *Neurology*. Dec 10;81(24):e178-81. doi: 10.1212/01.wnl.0000437294.20817.99

Leen WG, Taher M, Verbeek MM, Kamsteeg EJ, van de Warrenburg BP, Willemsen MA (2014). GLUT1 deficiency syndrome into adulthood: a follow-up study. *Journal of Neurology*. Mar;261(3):589-99. doi: 10.1007/s00415-014-7240-z. <https://link.springer.com/article/10.1007/s00415-014-7240-z>

Leone A, De Amicis R, Lessa C, Tagliabue A, Trentani C, Ferraris C, Battezzati A, Veggiotti P, Foppiani A, Ravella S, Bertoli S (2019). Food and Food Products on the Italian Market for Ketogenic Dietary Treatment of Neurological Diseases. *Nutrients*. May 17;11(5):1104. doi: 10.3390/nu11051104. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6566354/pdf/nutrients-11-01104.pdf>

Marin-Valencia I, Good LB, Ma Q, et al (2013). Heptanoate as a neural fuel: energetic and neurotransmitter precursors in normal and glucose transporter I-deficient (G1D) brain. *Journal of Cerebral Blood Flow Metabolism*. Feb;33(2):175-82. doi: 10.1038/jcbfm.2012.151. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3564188/pdf/jcbfm2012151a.pdf>

Nakamura S, Muramatsu SI, Takino N, Ito M, Jimbo EF, Shimazaki K, Onaka T, Ohtsuki S, Terasaki T, Yamagata T, Osaka H (2018). Gene therapy for Glut1-deficient mouse using an adeno-associated virus vector with the human intrinsic GLUT1 promoter. *J Gene Med*. Apr;20(4):e3013. doi: 10.1002/jgm.3013.

Oguni H, Ito Y, Otani Y, Nagata S (2018). Questionnaire survey on the current status of ketogenic diet therapy in patients with glucose transporter 1 deficiency syndrome (GLUT1DS) in Japan. *European Journal of Paediatric Neurology*. May;22(3):482-487. doi: 10.1016/j.ejpn.2017.12.013.

Opitz JM, Holt MC (1990). Microcephaly: general considerations and aids to nosology. *J Craniofac Genet Dev Biol*;10(2):175-204. PMID: 2211965

Pearson TS, Akman C, Hinton VJ, Engelstad K, De Vivo DC (2013). Phenotypic spectrum of glucose transporter type 1 deficiency syndrome (Glut1 DS). *Curr Neurol Neurosci Rep*. Apr;13(4):342. doi: 10.1007/s11910-013-0342-7.

Pearson TS, Pons R, Engelstad K, Kane SA, Goldberg ME, De Vivo DC (2017). Paroxysmal eye-head movements in Glut1 deficiency syndrome. *Neurology*. Apr 25;88(17):1666-1673. doi: 10.1212/WNL.0000000000003867. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5405761/pdf/NEUROLOGY2016766170.pdf>

Pong AW, Geary B, Engelstad KM, Natarajan A, Yang H, De Vivo DC (2012). Glucose transporter type I deficiency syndrome: epilepsy phenotypes and outcomes. *Epilepsia*. Sep;53(9):1503-10. doi: 10.1111/j.1528-1167.2012.03592.x. <https://onlinelibrary.wiley.com/doi/epdf/10.1111/j.1528-1167.2012.03592.x>

Read AP, Donnai D (2012). What can be offered to couples at (possibly) increased genetic risk? *J Community Genet*. Jul;3(3):167-74. doi: 10.1007/s12687-012-0105-1. [https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3419288/pdf/12687\\_2012\\_Article\\_105.pdf](https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3419288/pdf/12687_2012_Article_105.pdf)

Tang M, Park SH, De Vivo DC, Monani UR (2019). Therapeutic strategies for glucose transporter 1 deficiency syndrome. *Ann Clin Transl Neurol*. 2019 Sep;6(9):1923-1932. doi: 10.1002/acn3.50881. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6764625/pdf/ACN3-6-1923.pdf>

Varesio C, Pasca L, Parravicini S, Zanaboni MP, Ballante E, Masnada S, Ferraris C, Bertoli S, Tagliabue A, Veggiotti P, De Giorgis V (2019). Quality of Life in Chronic Ketogenic Diet Treatment: The GLUT1DS Population Perspective. *Nutrients*. Jul 19;11(7):1650. doi: 10.3390/nu11071650. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6682968/pdf/nutrients-11-01650.pdf>

Veggiotti P, Burlina A, Coppola G, Cusmai R, De Giorgis V, Guerrini R, Tagliabue A, Dalla Bernardina B (2011). The ketogenic diet for Dravet syndrome and other epileptic encephalopathies: an Italian consensus. *Epilepsia*. Apr;52 Suppl 2:83-9. doi: 10.1111/j.1528-1167.2011.03010.x. <https://onlinelibrary.wiley.com/doi/epdf/10.1111/j.1528-1167.2011.03010.x>

Veggiotti P, De Giorgis V (2014). Dietary Treatments and New Therapeutic Perspective in GLUT1 Deficiency Syndrome. *Curr Treat Options Neurol.* May;16(5):291. doi: 10.1007/s11940-014-0291-8.

Wang D, Pascual JM, De Vivo D. Glucose Transporter Type 1 Deficiency Syndrome. 2002 Jul 30 [updated 2018 Mar 1]. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Stephens K, Amemiya A, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993–2020.

Willemsen MA, Vissers LE, Verbeek MM, van Bon BW, Geuer S, Gilissen C, Klepper J, Kwint MP, Leen WG, Pennings M, Wevers RA, Veltman JA, Kamsteeg EJ (2017). Upstream SLC2A1 translation initiation causes GLUT1 deficiency syndrome. *Eur J Hum Genet.* Jun;25(6):771-774. doi: 10.1038/ejhg.2017.45.  
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5477372/pdf/ejhg201745a.pdf>

Winczewska-Wiktor A, Hoffman-Zacharska D, Starczewska M, Kaczmarek I, Badura-Stronka M, Steinborn B (2020). Variety of symptoms of GLUT1 deficiency syndrome in three-generation family. *Epilepsy Behav.* May;106:107036. doi: 10.1016/j.yebeh.2020.107036.

### **Ketogenic Diet**

EPIGRAPH VOL. 22 ISSUE 5, FALL 2020: [Food for thought: The ketogenic diet as epilepsy treatment](#)