UNIVERSITAS GADJAH MADA
HUBUNGAN GENOTIP DAN FENOTIP DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY PADA ANAK DENGAN METODE PEMERIKSAAN NEXT-GENERATION SEQUENCING
Agung Triono, Prof. Dr. dr. Elisabeth Siti Herini, Sp.A(K); Prof. dr. Gunadi, PhD, Sp.BA, Subsp. DA(K)
Universitas Gadjah Mada, 2025 | Diunduh dari http://etd.repository.ugm.ac.id/

DAFTAR PUSTAKA

(UK), N.G.C., 2022, Effectiveness of genetic testing in determining the aetiology of epilepsy, *Effectiveness of genetic testing in determining the aetiology of epilepsy: Epilepsies in children, young people and adults: Evidence review C*. *https://www.ncbi.nlm.nih.gov/books/NBK586316/*, diakses 26 January 2025.

Absoud, M., Parr, J.R., Halliday, D., Pretorius, P., Zaiwalla, Z. & Jayawant, S., 2010, A novel ARX phenotype: Rapid neurodegeneration with Ohtahara syndrome and a dyskinetic movement disorder, Dev Med Child Neurol, 52, 3, 305–307.

Aimiuwu, O. V, 2021, Modeling Gene Therapy for Intractable Developmental and Epileptic Encephalopathy,

Allen, A.S., Berkovic, S.F., Cossette, P., Delanty, N., Dlugos, D., Winawer, M.R., 2013, De novo mutations in epileptic encephalopathies, Nature, 501, 7466, 217–221. http://dx.doi.org/10.1038/nature12439,.

Álvarez-Mora, M.I., Sánchez, A., Rodríguez-Revenga, L., Corominas, J., Rabionet, R., Puig, S. & Madrigal, I., 2022, Diagnostic yield of next-generation sequencing in 87 families with neurodevelopmental disorders, Orphanet J. Rare Dis. 17, 1, 1–10.

Amador, A., Bostick, C.D., Olson, H., Peters, J., Camp, C.R., Krizay, D., et al, 2020, Modelling and treating GRIN2A developmental and epileptic encephalopathy in mice, Brain, 143, 7, 2039–2057.

Ambrosino, P., Soldovieri, M.V., Bast, T., Turnpenny, P.D., Uhrig, S., Biskup, S., et al, 2018, De novo gain-of-function variants in KCNT2 as a novel cause of developmental and epileptic encephalopathy, Ann. Neurol, 83, 6, 1198–1204.

Asadi-Pooya, A.A. & Katibeh, P., 2023, Lennox Gastaut Syndrome, *Comprehensive Pharmacology*, 3, 105–122. *https://www.ncbi.nlm.nih.gov/books/NBK532965/*, diakses 27 January 2025.

Bahi-Buisson, N., Nectoux, J., Rosas-Vargas, H., Milh, M., Boddaert, N., Girard, B., et al, 2008, Key clinical features to identify girls with CDKL5 mutations, Brain, 131, 10, 2647–2661.

Barcia, G., Chemaly, N., Kuchenbuch, M., Eisermann, M., Gobin-Limballe, S., Ciorna, V., et al, 2019, Epilepsy with migrating focal seizures, Neurol. Genet, 5, 6, e363.

Barić, I., Staufner, C., Augoustides-Savvopoulou, P., Chien, Y.H., Dobbelaere, D., Grünert, S.C., et al, 2016, Consensus recommendations for the diagnosis, treatment and follow-up of inherited methylation disorders, *Journal of Inherited Metabolic Disease*, 40, 1, 5. *https://pmc.ncbi.nlm.nih.gov/articles/PMC5203850/*, diakses 26 January 2025.

Baumgartner, M.R., Hörster, F., Dionisi-Vici, C., Haliloglu, G., Karall, D., Chapman, K.A., et al, 2014, Proposed guidelines for the diagnosis and management of methylmalonic and propionic acidemia, *Orphanet Journal of Rare Diseases*, 9, 1, 1–36. *https://ojrd.biomedcentral.com/articles/10.1186/s13023-014-0130-8*, diakses 26 January 2025.

UNIVERSITAS GADJAH MADA
HUBUNGAN GENOTIP DAN FENOTIP DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY PADA ANAK DENGAN METODE PEMERIKSAAN NEXT-GENERATION SEQUENCING
Agung Triono, Prof. Dr. dr. Elisabeth Siti Herini, Sp.A(K); Prof. dr. Gunadi, PhD, Sp.BA, Subsp. DA(K)
Universitas Gadjah Mada, 2025 | Diunduh dari http://etd.repository.ugm.ac.id/

Beal, J.C., Cherian, K. & Moshe, S.L., 2012, Early-Onset Epileptic Encephalopathies: Ohtahara Syndrome and Early Myoclonic Encephalopathy, *Pediatric Neurology*, 47, 5, 317–323.

Berg, A.T., Mahida, S. & Poduri, A., 2021, KCNQ2 -DEE : developmental or epileptic encephalopathy ?, , 666–676.

Berkovic, S.F., Mulley, J.C., Scheffer, I.E. & Petrou, S., 2006, Human epilepsies: interaction of genetic and acquired factors, Trends Neurosci, 29, 7, 391–397.

Blueprint Genetics, 2016, A guide to understanding variant classification, https://submit.ncbi.nlm.nih.gov/ft/byid/nxpnxkpc/variant_classification_wp_vara41-03.pdf,.

Boison, D., 2012, Adenosine dysfunction in epilepsy, *Glia*, 60, 8, 1234–1243. *https://pubmed.ncbi.nlm.nih.gov/22700220/*, diakses 13 December 2024.

Boonsimma, P., Ittiwut, C., Kamolvisit, W., Ittiwut, R., Chetruengchai, W., Phokaew, C., et al, 2023, Exome sequencing as first-tier genetic testing in infantile-onset pharmacoresistant epilepsy: diagnostic yield and treatment impact, *European journal of human genetics : EJHG*, 31, 2, 179–187. *https://pubmed.ncbi.nlm.nih.gov/36198807/*, diakses 27 January 2025.

Bureau, M. & Bernardina, D., 2011, DRAVET SYNDROME Electroencephalographic characteristics of Dravet syndrome, , 52, 13–23.

Byrne, S., Enright, N. & Delanty, N., 2021, Precision therapy in the genetic epilepsies of childhood,

Carvill, G.L., Weckhuysen, S., McMahon, J.M., Hartmann, C., Møller, R.S., Hjalgrim, H., et al, 2014, GABRA1 and STXBP1: novel genetic causes of Dravet syndrome., Neurology, 82, 14, 1245–1253.

Cash, S.J., Mcgue, B.P., Reynolds, T.S. & Crist, E.R., 2020, PIGA related disorder as a range of phenotypes rather than two distinct subtypes, Brain Dev., 42, 2, 205–210. https://doi.org/10.1016/j.braindev.2019.10.002,.

Cetica, V., Chiari, S., Mei, D., Parrini, E., Grisotto, L., Marini, C., et al, 2017, Clinical and genetic factors predicting Dravet syndrome in infants with SCN1A mutations, Neurology, 88, 11, 1037–1044.

Clapp, P., Bhave, S. V & Hoffman, P.L., 2008, How adaptation of the brain to alcohol leads to dependence: a pharmacological perspective., Alcohol Res Health : the journal of the National Institute on Alcohol Abuse and Alcoholism, 31, 4, 310–39. http://www.ncbi.nlm.nih.gov/pubmed/20729980%0Ahttp://www.pubmedcentral.nih.gov/articlerender.fcgi?artid=PMC2923844,.

Clark, M.M., Stark, Z., Farnaes, L., Tan, T.Y., White, S.M., Dimmock, D., 2018, Meta-analysis of the diagnostic and clinical utility of genome and exome sequencing and chromosomal microarray in children with suspected genetic diseases, NPJ Genom. Med., 3, 1, 1–10. http://dx.doi.org/10.1038/s41525-018-0053-8,.

Colasante, G., Lignani, G., Brusco, S., Di Berardino, C., Carpenter, J., Giannelli, S., et al, 2020, dCas9-Based Scn1a Gene Activation Restores Inhibitory Interneuron Excitability and Attenuates Seizures in Dravet Syndrome Mice, Mol. Ther., 28, 1, 235–253. https://www.sciencedirect.com/science/article/pii/S1525001619304010,.

UNIVERSITAS GADJAH MADA
HUBUNGAN GENOTIP DAN FENOTIP DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY PADA ANAK DENGAN METODE PEMERIKSAAN NEXT-GENERATION SEQUENCING
Agung Triono, Prof. Dr. dr. Elisabeth Siti Herini, Sp.A(K); Prof. dr. Gunadi, PhD, Sp.BA, Subsp. DA(K)
Universitas Gadjah Mada, 2025 | Diunduh dari http://etd.repository.ugm.ac.id/

Costain, G., Cordeiro, D., Matviychuk, D. & Mercimek-Andrews, S., 2019, Clinical Application of Targeted Next-Generation Sequencing Panels and Whole Exome Sequencing in Childhood Epilepsy, Neuroscience, 418, 291–310. https://doi.org/10.1016/j.neuroscience.2019.08.016,.

D'Gama, A.M., Mulhern, S., Sheidley, B.R., Boodhoo, F., Buts, S., Chandler, N.J., et al, 2023, Evaluation of the feasibility, diagnostic yield, and clinical utility of rapid genome sequencing in infantile epilepsy (Gene-STEPS): an international, multicentre, pilot cohort study, *The Lancet. Neurology*, 22, 9, 812–825. *https://pubmed.ncbi.nlm.nih.gov/37596007/*, diakses 27 January 2025.

Depienne, C., Gourfinkel-An, I., Baulac, S. & LeGuern, E., 2012, Genes in infantile epileptic encephalopathies,

Devaux, J., Dhifallah, S., De Maria, M., Stuart-Lopez, G., Becq, H., Milh, M., et al, 2017, A possible link between KCNQ2- and STXBP1-related encephalopathies: STXBP1 reduces the inhibitory impact of syntaxin-1A on M current, Epilepsia, 58, 12, 2073–2084.

Ding, J., Li, X., Tian, H., Wang, L., Guo, B., Wang, Y., et al, 2021, SCN1A Mutation—Beyond Dravet Syndrome: A Systematic Review and Narrative Synthesis, Front. Neurol., 12, December, 1–12.

Dunn, P., Albury, C.L., Maksemous, N., Benton, M.C., Sutherland, H.G., Smith, R.A., et al, 2018, Next generation sequencing methods for diagnosis of epilepsy syndromes, Front Genet, 9, FEB, 1–11.

Efthymiou, S., Salpietro, V., Bettencourt, C. & Houlden, H., 2018, Paroxysmal Movement Disorder and Epilepsy Caused by a De Novo Truncating Mutation in KAT6A, Journal of Pediatric Genetics, 07, 03, 114–116.

Ekşioğlu, Y.Z., Pong, A.W. & Takeoka, M., 2011, A novel mutation in the aristaless domain of the ARX gene leads to Ohtahara syndrome, global developmental delay, and ambiguous genitalia in males and neuropsychiatric disorders in females, Epilepsia, 52, 5, 984–992.

Endele, S., Rosenberger, G., Geider, K., Popp, B., Tamer, C., et al, 2010, Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes, Nature Publishing Group, 42, 11, 1021–1026. http://dx.doi.org/10.1038/ng.677,.

Esmaeeli Nieh, S. & Sherr, E.H., 2014, Epileptic encephalopathies: new genes and new pathways, Neurotherapeutics, 11, 4, 796–806.

Fisher, R.S., Acevedo, C., Arzimanoglou, A., Bogacz, A., Cross, J.H., Elger, C.E., et al, 2014, ILAE Official Report: A practical clinical definition of epilepsy, Epilepsia, 55, 4, 475–482.

Fisher, R.S., Cross, J.H., D'Souza, C., French, J.A., Haut, S.R., Higurashi, N., et al, 2017, Instruction manual for the ILAE 2017 operational classification of seizure types, Epilepsia, 58, 4, 531–542.

Fogel, B.L., Satya-Murti, S. & Cohen, B.H., 2016, Clinical exome sequencing in neurologic disease, *Neurology: Clinical Practice*, 6, 2, 164–176.

Forman, E.B., Gorman, K.M., Conroy, J., Arthur, N., Grant, C., Ennis, S., et al, 2018, Cost of exome sequencing in epileptic encephalopathy: Is it worth it'?, Arch. Dis. Child., 103, 3, 304.

UNIVERSITAS GADJAH MADA
HUBUNGAN GENOTIP DAN FENOTIP DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY PADA ANAK DENGAN METODE PEMERIKSAAN NEXT-GENERATION SEQUENCING
Agung Triono, Prof. Dr. dr. Elisabeth Siti Herini, Sp.A(K); Prof. dr. Gunadi, PhD, Sp.BA, Subsp. DA(K)
Universitas Gadjah Mada, 2025 | Diunduh dari http://etd.repository.ugm.ac.id/

Friocourt, G. & Parnavelas, J.G., 2010, Mutations in ARX result in several defects involving GABAergic neurons, Front. Cell. Neurosci., 4, MAR, 1–11.

Gertler, T., Bearden, D., Bhattacharjee, A. & Carvill, G., 2020, KCNT1 -Related Epilepsy Summary Genetic counseling, , 1–19.

Goetz, L.H., Schork, N.J., Clinic, S., Group, M., Jolla, L., Craig, J., et al, 2019, Personalized Medicine: Motivation, Challenges and Progress Laura, , 109, 6, 952–963.

Gomes, A. & Korf, M.S.B., 2018, Genet. Test, Elsevier Inc. http://dx.doi.org/10.1016/B978-0-323-48555-5.00005-3,.

Gong, P., Xue, J., Jiao, X., Zhang, Y. & Yang, Z., 2021, Genetic Etiologies in Developmental and / or Epileptic Encephalopathy With Electrical Status Epilepticus During Sleep : Cohort Study, , 12, April.

González, H.F.J., Yengo-kahn, A., Englot, D.J., Sciences, R. & Stimulator, V.N., 2020, Vagus Nerve Stimulation for the Treatment of Epilepsy Hernán, , 30, 2, 219–230.

Goto, A., Ishii, A., Shibata, M., Ihara, Y., Cooper, E.C. & Hirose, S., 2019, Characteristics of KCNQ2 variants causing either benign neonatal epilepsy or developmental and epileptic encephalopathy, January, 1–11.

Goto, A., Ishii, A., Shibata, M., Ihara, Y., Cooper, E.C. & Hirose, S., 2019, Characteristics of KCNQ2 variants causing either benign neonatal epilepsy or developmental and epileptic encephalopathy, Epilepsia, 60, 9, 1870–1880. https://pubmed.ncbi.nlm.nih.gov/31418850/, diakses 6 September 2024.

Guerrini, R., 2006, Epilepsy in children, Lancet, 367, 9509, 499–524.

Guerrini, R., Striano, P., Catarino, C. & Sisodiya, S.M., 2011, Neuroimaging and neuropathology of Dravet syndrome, Epilepsia, 52, SUPPL. 2, 30–34.

Gürsoy, S. & Erçal, D., 2016, Diagnostic Approach to Genetic Causes of Early-Onset Epileptic Encephalopathy, J. Child Neurol., 31, 4, 523–532.

Hagemann, I.S., 2015, Overview of Technical Aspects and Chemistries of Next-Generation Sequencing, Elsevier Inc. http://dx.doi.org/10.1016/B978-0-12-404748-8.00001-0,.

Hamdan, F.F. & Michaud, J.L., 2020, SYNGAP1 -Related Intellectual Disability Summary Genetic counseling, , 1–13.

Hamdan, F.F., Saitsu, H., Nishiyama, K., Gauthier, J., Dobrzeniecka, S., Spiegelman, D., et al, 2012, Identification of a novel in-frame de novo mutation in SPTAN1 in intellectual disability and pontocerebellar atrophy, Eur. J. Hum. Genet., 20, 7, 796–800.

Han, J.Y. & Lee, I.G., 2020, Genetic tests by next-generation sequencing in children with developmental delay and/or intellectual disability, Korean J. Pediatr, 63, 6, 195–202.

Han, Z., Chen, C., Christiansen, A., Ji, S., Lin, Q., Anumonwo, C., et al, 2020, Antisense oligonucleotides increase Scn1a expression and reduce seizures and SUDEP incidence in a mouse model of Dravet syndrome, Sci Transl Med., 12, 558.

Heather, J.M. & Chain, B., 2016, The sequence of sequencers: The history of sequencing DNA, Genomics, 107, 1, 1–8. http://dx.doi.org/10.1016/j.ygeno.2015.11.003,.

UNIVERSITAS GADJAH MADA
HUBUNGAN GENOTIP DAN FENOTIP DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY PADA ANAK DENGAN METODE PEMERIKSAAN NEXT-GENERATION SEQUENCING
Agung Triono, Prof. Dr. dr. Elisabeth Siti Herini, Sp.A(K); Prof. dr. Gunadi, PhD, Sp.BA, Subsp. DA(K)
Universitas Gadjah Mada, 2025 | Diunduh dari http://etd.repository.ugm.ac.id/

Helbig, K.L., Farwell Hagman, K.D., Shinde, D.N., Mroske, C., Powis, Z., Li, S., et al, 2016, Diagnostic exome sequencing provides a molecular diagnosis for a significant proportion of patients with epilepsy, Genet. Med , 18, 9, 898–905. https://www.sciencedirect.com/science/article/pii/S1098360021044348,.

Hernandez, A. & Wirrel, E., 2019, Infantile Spasms | West Syndrome | Epilepsy Foundation, https://www.epilepsy.com/what-is-epilepsy/syndromes/infantile-spasms-west-syndrome, diakses 28 August 2022.

Hernandez, A. & Wirrell, E., 2019, Infantile Spasms | West Syndrome | Epilepsy Foundation, https://www.epilepsy.com/what-is-epilepsy/syndromes/infantile-spasms-west-syndrome, diakses 3 September 2022.

Hernandez, A. & Wirrell, E., 2020, Epilepsy of Infancy with Migrating Focal Seizures | Epilepsy Foundation, https://www.epilepsy.com/what-is-epilepsy/syndromes/epilepsy-infancy-migrating-focal-seizures, diakses 3 September 2022.

Hernandez, A., Kiriakopoulos, E. & Wirrell, E., 2019a, Ohtahara Syndrome | Epilepsy Foundation, https://www.epilepsy.com/what-is-epilepsy/syndromes/ohtahara-syndrome, diakses 3 September 2022.

Hernandez, A., Kiriakopoulos, E. & Wirrell, E., 2019b, Ohtahara Syndrome | Epilepsy Foundation, https://www.epilepsy.com/what-is-epilepsy/syndromes/ohtahara-syndrome, diakses 28 August 2022.

Hwang, S.K. & Kwon, S., 2015, Early-onset epileptic encephalopathies and the diagnostic approach to underlying causes, Korean J. Ped., 58, 11, 407–414.

Iskandar, K., Herini, E.S., Triono, A., Hadiyanto, M.L., Nugrahanto, A.P. & Gunadi, 2023, Phenotypes of a female patient with novel de novo frameshift ARX variant identified by whole-exome sequencing: a case report, *Annals of Medicine and Surgery*, 85, 2, 236–241. *https://journals.lww.com/annals-of-medicine-and-surgery/fulltext/2023/02000/phenotypes_of_a_female_patient_with_novel_de_novo.35.aspx*, diakses 19 December 2024.

Jain, P., Sharma, S. & Tripathi, M., 2013, Diagnosis and management of epileptic encephalopathies in children, Epilepsy Res, 2013.

Jdila, M. Ben, Issa, A. Ben, Khabou, B., Rhouma, et al, 2017, Novel mutations in the CDKL5 gene in complex genotypes associated with West syndrome with variable phenotype: First description of somatic mosaic state, Clin. Chim. Acta, 473, 51–59. http://dx.doi.org/10.1016/j.cca.2017.08.001,.

Joseph, D.J., Deimling, M. Von, Hasegawa, Y., Risbud, R., Mccoy, A.J. & Marsh, E.D., 2021, Postnatal Arx transcriptional activity regulates functional properties of PV interneurons iScience Postnatal Arx transcriptional activity regulates functional properties, ISCIENCE, 24, 1, 101999. https://doi.org/10.1016/j.isci.2020.101999,.

Joshi, C. & Wirrell, E., 2020a, Dravet Syndrome | Epilepsy Foundation, https://www.epilepsy.com/what-is-epilepsy/syndromes/dravet-syndrome, diakses 28 August 2022.

Joshi, C. & Wirrell, E., 2020b, Early Myoclonic Encephalopathy (EME) | Epilepsy Foundation, https://www.epilepsy.com/what-is-epilepsy/syndromes/early-myoclonic-encephalopathy, diakses 3 September 2022.

UNIVERSITAS GADJAH MADA
HUBUNGAN GENOTIP DAN FENOTIP DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY PADA ANAK DENGAN METODE PEMERIKSAAN NEXT-GENERATION SEQUENCING
Agung Triono, Prof. Dr. dr. Elisabeth Siti Herini, Sp.A(K); Prof. dr. Gunadi, PhD, Sp.BA, Subsp. DA(K)
Universitas Gadjah Mada, 2025 | Diunduh dari http://etd.repository.ugm.ac.id/

Joshi, C., Ciliberto, M. & Wirrell, E., 2020, Epileptic Encephalopathy Continuous Spike and Wave During Sleep CSWS | Epilepsy Foundation, https://www.epilepsy.com/what-is-epilepsy/syndromes/epileptic-encephalopathy-continuous-spike-and-wave-during-sleep, diakses 28 August 2022.

Joshi, C., Kolbe, D.L., Mansilla, M.A., Mason, S., Smith, R.J.H. & Campbell, C.A., 2016, Ketogenic diet - A novel treatment for early epileptic encephalopathy due to PIGA deficiency., Brain Dev., 38, 9, 848–851.

Kato, M., 2006, A new paradigm for West syndrome based on molecular and cell biology, Epilepsy Res., 70, SUPPL.1, 87–95.

Kato, M., Das, S., Petras, K., Kitamura, K., Morohashi, K.I., Abuelo, D.N., et al, , 2004, Mutations of ARX Are Associated with Striking Pleiotropy and Consistent Genotype-Phenotype Correlation, Hum. Mutat., 23, 2, 147–159.

Kato, M., Saitsu, H., Murakami, Y., Kikuchi, K., Watanabe, S., et al, 2014, PIGA mutations cause early-onset epileptic encephalopathies and distinctive features., Neurology, 82, 18, 1587–1596.

Khan, S. & Al Baradie, R., 2012, Epileptic Encephalopathies: An Overview, Epilepsy Res Treat., 2012, 1–8.

Kilstrup-Nielsen, C., Rusconi, L., La Montanara, P., Ciceri, D., Bergo, A., Bedogni, 2012, What we know and would like to know about CDKL5 and its involvement in epileptic encephalopathy, Neural Plasticity, 2012.

Kim, J.H., Yang, D., Kim, S.H., Won, D., HeungDong, K., Soo, J., et al, 2020, Clinical characteristics of KCNQ2 encephalopathy, Brain and Development, 2–8. https://doi.org/10.1016/j.braindev.2020.08.015,.

Kim, J.H., Yang, D., Kim, S.H., Won, D., HeungDong, K., Soo, J., et al, 2020, Clinical characteristics of KCNQ2 encephalopathy, *Brain and Development*, 2–8. *https://doi.org/10.1016/j.braindev.2020.08.015*,.

Kiriakopoulos, E., Wirrell, E., Sirven, J.I. & Shafer, P.O., 2019, Case Study on Lennox-Gastaut Syndrome | Epilepsy Foundation, https://www.epilepsy.com/what-is-epilepsy/syndromes/lennox-gastaut-syndrome, diakses 28 August 2022.

Kosaki, K., Udaka, T. & Okuyama, T., 2005, DHPLC in clinical molecular diagnostic services, *Molecular Genetics and Metabolism*, 86, 1–2, 117–123.

Kothur, K., Holman, K., Farnsworth, E., Ho, G., Lorentzos, M., Troedson, C., et al, 2018, Diagnostic yield of targeted massively parallel sequencing in children with epileptic encephalopathy., Seizure, 59, 132–140.

Kural, Z. & Ozer, A.F., 2012, Epileptic Encephalopathies in Adults and Childhood, Epilepsy Res Treat., 2012, 1–8.

Lee, Jiwon, Lee, C., Ki, C.-S. & Lee, Jeehun, 2020, Determining the best candidates for next-generation sequencing-base gene panel for evaluation of early-onset epilepsy., Mol. Genet. Genomic Med., 8, 9, e1376.

Lemke, J.R., Lal, D., Reinthaler, E.M., Steiner, I., Nothnagel, M., … Von Spiczak, S., 2013, Mutations in GRIN2A cause idiopathic focal epilepsy with rolandic spikes, Nat. Genet., 45, 9, 1067–1072. http://dx.doi.org/10.1038/ng.2728,.

Lesca, G., Møller, R.S., Rudolf, G., Hirsch, E., Hjalgrim, H. & Szepetowski, P., 2019, Update on the genetics of the epilepsy-aphasia spectrum and role of

UNIVERSITAS GADJAH MADA
HUBUNGAN GENOTIP DAN FENOTIP DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY PADA ANAK DENGAN METODE PEMERIKSAAN NEXT-GENERATION SEQUENCING
Agung Triono, Prof. Dr. dr. Elisabeth Siti Herini, Sp.A(K); Prof. dr. Gunadi, PhD, Sp.BA, Subsp. DA(K)
Universitas Gadjah Mada, 2025 | Diunduh dari http://etd.repository.ugm.ac.id/

GRIN2A mutations., Epileptic Disord. : international epilepsy journal with videotape, 21, S1, 41–47.

Lesca, G., Rudolf, G., Bruneau, N., Lozovaya, N., Labalme, A., Boutry-kryza, N., et al, 2013, GRIN2A mutations in acquired epileptic aphasia and related childhood focal epilepsies and encephalopathies with speech and language dysfunction, Nat. Genet., , July, 1–8. http://dx.doi.org/10.1038/ng.2726,.

Lesca, G., Rudolf, G., Bruneau, N., Lozovaya, N., Labalme, A., Boutry-Kryza, N., et al, 2013, GRIN2A mutations in acquired epileptic aphasia and related childhood focal epilepsies and encephalopathies with speech and language dysfunction, Nat. Genet., 45, 9, 1061–1066. http://dx.doi.org/10.1038/ng.2726,.

Li, T., Cheng, M., Wang, J., Hong, S., Li, M., Liao, S., et al, 2018, De novo mutations of STXBP1 in Chinese children with early onset epileptic encephalopathy., Genes Brain Behav., 17, 8, e12492.

Liang, J.-S., Huang, H., Wang, J.-S. & Lu, J.-F., 2019, Phenotypic manifestations between male and female children with CDKL5 mutations., Brain Dev., 41, 9, 783–789.

Liu, S., Wang, L., Cai, X.T., Zhou, H., Yu, D. & Wang, Z., 2008, Therapeutic bene fi ts of ACTH and levetiracetam in STXBP1 encephalopathy with a de novo mutation,

Liu, W., Smith, D.I., Rechtzigel, K.J., Thibodeau, S.N. & James, C.D., 1998, Denaturing high performance liquid chromatography (DHPLC) used in the detection of germline and somatic mutations, *Nucleic Acids Research*, 26, 6, 1396–1400. *https://dx.doi.org/10.1093/nar/26.6.1396*, diakses 28 January 2025.

Low, K.J., James, M., Sharples, P.M., Eaton, M., Jenkinson, S.,., 2018, A novel PIGA variant associated with severe X-linked epilepsy and profound developmental delay, Seizure, 56, 1–3. https://doi.org/10.1016/j.seizure.2018.01.013,.

Mao, X., Bruneau, N., Gao, Q., Becq, H., Jia, Z., Xi, H., et al, 2020, The Epilepsy of Infancy With Migrating Focal Seizures: Identification of de novo Mutations of the KCNT2 Gene That Exert Inhibitory Effects on the Corresponding Heteromeric KNa1.1/KNa1.2 Potassium Channel, Front. Cell. Neurosci, 14, January, 1–12.

Marsh, E.D. & Golden, J.A., 2013, Developing Models of Aristaless-Related Homeobox Mutations, Jasper's Basic Mechanisms of the Epilepsies, , Md, 813–823.

Mastrangelo, M. & Leuzzi, V., 2012, Genes of early-onset epileptic encephalopathies: From genotype to phenotype, Ped Neurol, 46, 1, 24–31. http://dx.doi.org/10.1016/j.pediatrneurol.2011.11.003,.

McTague, A. & Cross, J.H., 2013, Treatment of epileptic encephalopathies, CNS Drugs, 27, 3, 175–184.

McTague, A., Howell, K.B., Cross, J.H., Kurian, M.A. & Scheffer, I.E., 2016, The genetic landscape of the epileptic encephalopathies of infancy and childhood, The Lancet Neurol, 15, 3, 304–316. http://dx.doi.org/10.1016/S1474-4422(15)00250-1,.

UNIVERSITAS GADJAH MADA
HUBUNGAN GENOTIP DAN FENOTIP DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY PADA ANAK DENGAN METODE PEMERIKSAAN NEXT-GENERATION SEQUENCING
Agung Triono, Prof. Dr. dr. Elisabeth Siti Herini, Sp.A(K); Prof. dr. Gunadi, PhD, Sp.BA, Subsp. DA(K)
Universitas Gadjah Mada, 2025 | Diunduh dari http://etd.repository.ugm.ac.id/

Mei, D., Cetica, V., Marini, C. & Guerrini, R., 2019, Dravet syndrome as part of the clinical and genetic spectrum of sodium channel epilepsies and encephalopathies, Epilepsia, 60, S3, S2–S7.

Mercimek-mahmutoglu, S., Patel, J., Cordeiro, D., Hewson, S., Callen, D., Donner, et al, 2015, Diagnostic yield of genetic testing in epileptic encephalopathy in childhood, , 707–716.

Miceli, F., Soldovieri, M.V., Weckhuysen, S., Cooper, E. & Taglialatela, M., 2022, KCNQ2 -Related Disorders, , 1–27.

Mignot, C., von Stülpnage, C., Nava, C., Ville, D., Sanlaville, D., … von Spiczak, S., 2016, Genetic and neurodevelopmental spectrum of SYNGAP1-associated intellectual disability and epilepsy, J. Med. Genet., 53, 8, 511–522.

Millichap, J.J., Park, K.L., Tsuchida, T., Carmant, L., Flamini, R., Levisohn, P.M.,et al., 2016, KCNQ2 encephalopathy, , 0, 1–9.

Morrow, J.S. & Stankewich, M.C., 2021, The Spread of Spectrin in Ataxia and Neurodegenerative Disease, J. Exp. Neurol, 2, 3, 131–139.

Mulcahey, P.J., Tang, S., Takano, H., White, A., Davila Portillo, D.R., Kane, O.M., Marsh, E.D., Zhou, Z. & Coulter, D.A., 2020, Aged heterozygous Cdkl5 mutant mice exhibit spontaneous epileptic spasms, Exp. Neurol., 332, 1–21.

Myers, C.T., McMahon, J.M., Schneider, A.L., Petrovski, S., Allen, A.S., et al, 2016, De Novo Mutations in SLC1A2 and CACNA1A Are Important Causes of Epileptic Encephalopathies, Am. J. Hum. Genet., 99, 2, 287–298.

Myers, K.A. & Scheffer, I.E., 2019, GRIN2A -Related Speech Disorders and Epilepsy Summary Genetic counseling Suggestive Findings, , 1–16.

Myers, S.J., Yuan, H., Kang, J.Q., Tan, F.C.K., Traynelis, S.F. & Low, C.M., 2019, Distinct roles of GRIN2A and GRIN2B variants in neurological conditions [version 1; peer review: 2 approved], F1000Research, 8, 1–14.

Nishimura, M., Takano, A., Mizuguchi, T., Saitsu, H., Takeuchi, Y. & Matsumoto, N., 2008, CDKL5 disruption by t(X;18) in a girl with West syndrome, Clin. Genet., 74, 3, 288–290.

Nonoda, Y., Saito, Y., Nagai, S., Sasaki, M., Iwasaki, T., Matsumoto, N., 2013, Progressive diffuse brain atrophy in West syndrome with marked hypomyelination due to SPTAN1 gene mutation, Brain Dev., 35, 3, 280–283. http://dx.doi.org/10.1016/j.braindev.2012.05.002,.

Oguni, H., 2010, What is Lennox-Gastaut syndrome in the modern era?, *Neurology Asia*, 15, 9–10.

Olson, H.E., Kelly, M., LaCoursiere, C.M., Pinsky, R., Tambunan, D., Shain, C.,et al., 2017, Genetics and genotype–phenotype correlations in early onset epileptic encephalopathy with burst suppression, Ann. Neurol., 81, 3, 419–429.

Olson, Heather E. Kelly, McKenna. Pinsky, R., Tambunan, D., et al, 2018, Genetics and genotype-phenotype correlations in early onset epileptic encephalopathy with burst suppression, HHS Public Access, 81, 3, 419–429.

Patel, J. & Mercimek-Mahmutoglu, S., 2016, Epileptic Encephalopathy in Childhood: A Stepwise Approach for Identification of Underlying Genetic Causes, Indian J. Pediatr., 83, 10, 1164–1174.

Perucca, P., Scheffer, I.E. & Kiley, M., 2017, The management of epilepsy in children and adults,

UNIVERSITAS GADJAH MADA
HUBUNGAN GENOTIP DAN FENOTIP DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY PADA ANAK DENGAN METODE PEMERIKSAAN NEXT-GENERATION SEQUENCING
Agung Triono, Prof. Dr. dr. Elisabeth Siti Herini, Sp.A(K); Prof. dr. Gunadi, PhD, Sp.BA, Subsp. DA(K)
Universitas Gadjah Mada, 2025 | Diunduh dari http://etd.repository.ugm.ac.id/

Pierson, T.M., Yuan, H., Marsh, E.D., Fuentes-Fajardo, K., Adams, D.R., Markello, T., et al, 2014, GRIN2A mutation and early-onset epileptic encephalopathy: personalized therapy with memantine, Ann. Clin. Transl. Neurol., 1, 3, 190–198.

Rampazzo, A.C.M., dos Santos, R.R.P., Maluf, F.A., Simm, R.F., Marson, F.A.L.,., 2021, Dravet syndrome and Dravet syndrome-like phenotype: a systematic review of the SCN1A and PCDH19 variants, Neurogenetics, 22, 2, 105–115. https://doi.org/10.1007/s10048-021-00644-7,.

Rapaccini, V., Esposito, S., Strinati, F., Allegretti, M., Manfroi, E., Miconi, F.,et al., 2018, A child with a c.6923_6928dup (p.Arg2308_Met2309dup) SPTAN1 mutation associated with a severe early infantile epileptic encephalopathy, Int. J. Mol. Sci., 19, 7, 1–6.

Ricci, R. & Colasante, G., 2021, CRISPR / dCas9 as a Therapeutic Approach for Neurodevelopmental Disorders : Innovations and Limitations Compared to Traditional Strategies, , 253–261.

Richards, C.S., Aziz, N., Bale, S., Bick, D., Das, S. & Gastier-Foster, J., 2015, Standards and guidelines for the interpretation of sequence variants : A Joint Consensus Recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology, HHS Public Access, 96, S261, 134–134.

Riney, K., Bogacz, A., Somerville, E. & Hirsch, E., 2022, International League Against Epilepsy classification and definition of epilepsy syndromes with onset at a variable age : position statement by the ILAE Task Force on Nosology and Definitions, April 2021, 1443–1474.

Russo, A. & Gobbi, G., 2020, A reflection on the role of genetics in the concept of "epileptic encephalopathy", as emerged from the most recent ILEA classification of epilepsy, Ital. J. Pediatr., 46, 1, 1–5.

Saitsu, H., Kato, M. & Matsumoto, N., 2010, Haploinsufficiency of STXBP1 and Ohtahara syndrome, Epilepsia, 51, SUPPL. 5, 71.

Saitsu, H., Tohyama, J., Kumada, T., Egawa, K., Hamada, K., Okada, I., et al, 2010, Dominant-Negative Mutations in α-II Spectrin Cause West Syndrome with Severe Cerebral Hypomyelination, Spastic Quadriplegia, and Developmental Delay, iAm. J. Hum. Genet, 86, 6, 881–891. http://dx.doi.org/10.1016/j.ajhg.2010.04.013,.

Santoro, C., Bernardo, P., Coppola, A., Pugliese, U., Cirillo, M., Giugliano, T., et al, 2018, Seizures in children with neurofibromatosis type 1: is neurofibromatosis type 1 enough?, Italian Journal of Pediatrics, 44, 1. /pmc/articles/PMC5863905/, diakses 2 September 2024.

Scheffer, I.E. & Liao, J., 2020, Deciphering the concepts behind "Epileptic encephalopathy" and "Developmental and epileptic encephalopathy," European Journal of Paediatric Neurology, 24, 11–14. https://doi.org/10.1016/j.ejpn.2019.12.023,.

Scheffer, I.E., Berkovic, S., Capovilla, G., Connolly, M.B., French, J., Guilhoto, L., et al, 2017, ILAE classification of the epilepsies: Position paper of the ILAE Commission for Classification and Terminology, Epilepsia, 58, 4, 512–521.

UNIVERSITAS GADJAH MADA
HUBUNGAN GENOTIP DAN FENOTIP DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY PADA ANAK DENGAN METODE PEMERIKSAAN NEXT-GENERATION SEQUENCING
Agung Triono, Prof. Dr. dr. Elisabeth Siti Herini, Sp.A(K); Prof. dr. Gunadi, PhD, Sp.BA, Subsp. DA(K)
Universitas Gadjah Mada, 2025 | Diunduh dari http://etd.repository.ugm.ac.id/

Seo, G.H., Kim, T., Choi, I.H., Park, J. young, Lee, J., Kim, S., et al, 2020, Diagnostic yield and clinical utility of whole exome sequencing using an automated variant prioritization system, EVIDENCE, *Clinical Genetics*, 98, 6, 562. *https://pmc.ncbi.nlm.nih.gov/articles/PMC7756481/*, diakses 28 January 2025.

Shao, L.R., Habela, C.W. & Stafstrom, C.E., 2019, Pediatric epilepsy mechanisms: Expanding the paradigm of excitation/inhibition imbalance, Children, 6, 2.

Sharma, S. & Tripathi, M., 2013, Ketogenic Diet in Epileptic Encephalopathies, Epilepsy Research and Treatment, 2013, 1–5.

Shoubridge, C., Jackson, M., Grinton, B., Berkovic, S.F., Scheffer, I.E., Huskins, S., et al, 2019, Splice variant in ARX leading to loss of C-terminal region in a boy with intellectual disability and infantile onset developmental and epileptic encephalopathy, Am. J. Hum. Genet., Part A, 179, 8, 1483–1490.

Sisodiya, S.M., 2021, Precision medicine and therapies of the future, 62, February 2020.

Specchio, N., Wirrell, E.C., Scheffer, I.E., Nabbout, R., Riney, K., Samia, P., et al , 2022, International League Against Epilepsy classification and definition of epilepsy syndromes with onset in childhood: Position paper by the ILAE Task Force on Nosology and Definitions, Epilepsia.

Stamberger, H., Nikanorova, M., Accorsi, P., Angriman, M., Benkel-herrenbrueck, I., Capovilla, et al, 2016, A neurodevelopmental disorder including epilepsy, Neurology, 86, 10, 954–962.

Steel, D., Symonds, J.D., Zuberi, S.M. & Brunklaus, A., 2017, Dravet syndrome and its mimics: Beyond SCN1A, Epilepsia, 58, 11, 1807–1816.

Strehlow, V., Heyne, H.O., Vlaskamp, D.R.M., Marwick, K.F.M., Rudolf, G., … Willemsen, M.H., 2019, GRIN2A -related disorders: Genotype and functional consequence predict phenotype, Brain, 142, 1, 80–92.

Symonds, J.D. & McTague, A., 2020, Epilepsy and developmental disorders: Next generation sequencing in the clinic, *European Journal of Paediatric Neurology*, 24, 15–23.

Syrbe, S., Harms, F.L., Parrini, E., Montomoli, M., Mütze, U., Helbig, K.L., et al, 2017, Delineating SPTAN1 associated phenotypes: From isolated epilepsy to encephalopathy with progressive brain atrophy, Brain, 140, 9, 2322–2336.

Tan, M.H., Gécz, J. & Shoubridge, C., 2013, PCR amplification and sequence analysis of GC-rich sequences: Aristaless-related homeobox example, Methods Mol. Biol., 1017, May, 105–120.

Tischfield, D.J., Saraswat, D.K., Furash, A., Fowler, S.C., Fuccillo, M. V. & Anderson, S.A., 2017, Loss of the neurodevelopmental gene Zswim6 alters striatal morphology and motor regulation, *Neurobiology of disease*, 103, 174. *https://pmc.ncbi.nlm.nih.gov/articles/PMC5703052/*, diakses 13 December 2024.

Triono, A., Herini, E.S. & Gunadi, 2024, Genetic exploration of Dravet syndrome: two case report, *Journal of Medical Case Reports*, 18, 1, 1–9. *https://jmedicalcasereports.biomedcentral.com/articles/10.1186/s13256-024-04514-2*, diakses 19 December 2024.

Triono, A., Herini, E.S., Iskandar, K., Hadiyanto, M.L., Diantika, K., Wijayanti, V.W., et al, 2023, De novo mutation of the TSC2 gene in patient with Tuberous

UNIVERSITAS GADJAH MADA
HUBUNGAN GENOTIP DAN FENOTIP DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY PADA ANAK DENGAN METODE PEMERIKSAAN NEXT-GENERATION SEQUENCING
Agung Triono, Prof. Dr. dr. Elisabeth Siti Herini, Sp.A(K); Prof. dr. Gunadi, PhD, Sp.BA, Subsp. DA(K)
Universitas Gadjah Mada, 2025 | Diunduh dari http://etd.repository.ugm.ac.id/

Sclerosis Complex-Associated Neuropsychiatric Disorders (TAND) Phenotype: a case report, *Annals of Medicine and Surgery*, 85, 5, 2102. *https://pmc.ncbi.nlm.nih.gov/articles/PMC10205250/*, diakses 18 December 2024.

Triono, A., Iskandar, K., Hadiyanto, M.L., Nugrahanto, A.P., Diantika, K., Wijayanti, V.W., et al, 2023, Identification of the genetic basis of pediatric neurogenetic disorders at a tertiary referral hospital in Indonesia: Contribution of whole exome sequencing, *PLOS ONE*, 18, 10, e0293113. *https://journals.plos.org/plosone/article?id=10.1371/journal.pone.0293113*, diakses 19 December 2024.

Trivisano, M. & Specchio, N., 2020, What are the epileptic encephalopathies?, Curr. Opin. Neurol., 33, 2, 179–184.

Turner, T.J., Zourray, C., Schorge, S. & Lignani, G., 2021, Recent advances in gene therapy for neurodevelopmental disorders with epilepsy, , , June 2020, 229–262.

van Dijk, E.L., Jaszczyszyn, Y., Naquin, D. & Thermes, C., 2018, The Third Revolution in Sequencing Technology, Trends Genet, 34, 9, 666–681. https://doi.org/10.1016/j.tig.2018.05.008,.

Vanlerberghe, C., Petit, F., Malan, V., Vincent-Delorme, C., Bouquillon, S., Boute, O., et al, 2015, 15q11.2 microdeletion (BP1-BP2) and developmental delay, behaviour issues, epilepsy and congenital heart disease: a series of 52 patients, *European journal of medical genetics*, 58, 3, 140–147. *https://pubmed.ncbi.nlm.nih.gov/25596525/*, diakses 13 December 2024.

Vlaskamp, D.R.M., Shaw, B.J., Burgess, R., Mei, D., Montomoli, M., Xie, H. et al, 2019, SYNGAP1 encephalopathy: A distinctive generalized developmental and epileptic encephalopathy, Neurology, 92, 2, E96–E107.

von Deimling, M., Helbig, I. & Marsh, E.D., 2017, Epileptic Encephalopathies—Clinical Syndromes and Pathophysiological Concepts, Curr Neurol Neurosci Rep ., 17, 2.

Wang, J., Lin, Z.J., Liu, L., Xu, H.Q., Shi, Y.W., Yi, Y.H., et al, 2017, Epilepsy-associated genes, Seizure, 44, 11–20.

Wang, Y., Ji, T., Nelson, A.D., Glanowska, K., Murphy, G.G., Jenkins, P.M. & Parent, J.M., 2018, Critical roles of αII spectrin in brain development and epileptic encephalopathy, J. Clin. Investig., 128, 2, 760–773.

Wheless, J.W., Fulton, S.P. & Mudigoudar, B.D., 2020, Dravet Syndrome: A Review of Current Management, Ped Neurol., 107, 28–40. https://doi.org/10.1016/j.pediatrneurol.2020.01.005,.

WHO, 2022, Epilepsy, https://www.who.int/news-room/fact-sheets/detail/epilepsy, diakses 27 August 2022.

Wimmer, V.C., Reid, C.A., So, E.Y., Berkovic, S.F. & Petrou, S., 2010, Axon initial segment dysfunction in epilepsy, , 11, 1829–1840.

Wirrell, E., Tinuper, P., Perucca, E. & Moshe, S.L., 2022, Introduction to the epilepsy syndrome papers, April, 1330–1332.

Wirrell, E.C., Nabbout, R., Scheffer, I.E., Alsaadi, T., Bogacz, A., French, J.A., et al, 2022, Methodology for classification and definition of epilepsy syndromes

UNIVERSITAS GADJAH MADA
HUBUNGAN GENOTIP DAN FENOTIP DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY PADA ANAK DENGAN METODE PEMERIKSAAN NEXT-GENERATION SEQUENCING
Agung Triono, Prof. Dr. dr. Elisabeth Siti Herini, Sp.A(K); Prof. dr. Gunadi, PhD, Sp.BA, Subsp. DA(K)
Universitas Gadjah Mada, 2025 | Diunduh dari http://etd.repository.ugm.ac.id/

with list of syndromes : Report of the ILAE Task Force on Nosology and Definitions, , , April 2021, 1333–1348.

Wong-Kisiel, L., 2022, Pyridoxine-Dependent Epilepsy – ALDH7A1, *Epilepsy Case Studies: Pearls for Patient Care*, 9783319013664, 7–11. *https://www.ncbi.nlm.nih.gov/books/NBK1486/*, diakses 26 January 2025.

Wu, Q., Wang, H., Fan, Y.Y., Zhang, J.M., Liu, X.Y., Fang, X.Y., et al, 2018a, Ketogenic diet effects on 52 children with pharmacoresistant epileptic encephalopathy: A clinical prospective study, Brain Behav., 8, 5, 1–8.

Wu, Q., Wang, H., Fan, Y.Y., Zhang, J.M., Liu, X.Y., Fang, X.Y., et al, 2018b, Ketogenic diet effects on 52 children with pharmacoresistant epileptic encephalopathy: A clinical prospective study, Brain Behav., 8, 5, 1–9.

Wykes, R.C. & Lignani, G., 2017, Gene therapy and editing: Novel potential treatments for neuronal channelopathies, Neuropharmacology. http://dx.doi.org/10.1016/j.neuropharm.2017.05.029,.

Xiao, W. & Oefner, P.J., 2001, Denaturing high-performance liquid chromatography: A review, *Human mutation*, 17, 6, 439–474. *https://pubmed.ncbi.nlm.nih.gov/11385705/*, diakses 28 January 2025.

Yamagata, T., Raveau, M., Kobayashi, K., Miyamoto, H., Tatsukawa, T., Ogiwara, I., et al, 2020, CRISPR/dCas9-based Scn1a gene activation in inhibitory neurons ameliorates epileptic and behavioral phenotypes of Dravet syndrome model mice, Neurobiol. Dis., 141, 104954. https://www.sciencedirect.com/science/article/pii/S0969996120302291,.

Yamatogi, Y. & Ohtahara, S., 2002, Early-infantile epileptic encephalopathy with suppression-bursts, Ohtahara syndrome; its overview referring to our 16 cases, *Brain and Development*, 24, 1, 13–23.

Zeng, F., Yang, Y., Xu, Z., Wang, Z., Ke, H., Zhang, J., et al, 2022, Clinical manifestations and genetic analysis of a newborn with Arboleda−Tham syndrome, *Frontiers in Genetics*, 13, 990098.

Zeng, L.H., Rensing, N.R., Zhang, B., Gutmann, D.H., Gambello, M.J. & Wong, M., 2011, Tsc2 gene inactivation causes a more severe epilepsy phenotype than Tsc1 inactivation in a mouse model of Tuberous Sclerosis Complex, Human Molecular Genetics, 20, 3, 445. /pmc/articles/PMC3016907/, diakses 27 August 2024.

Zhou, P., He, N., Zhang, J.W., Lin, Z.J., Wang, J., Yan, L.M., et al, 2018, Novel mutations and phenotypes of epilepsy-associated genes in epileptic encephalopathies, Genes Brain Behav., 17, 8.

Zuberi, S.M., Wirrell, E., Yozawitz, E., Wilmshurst, J.M., Specchio, N., Riney, K., et al, 2022, ILAE classification and definition of epilepsy syndromes with onset in neonates and infants: Position statement by the ILAE Task Force on Nosology and Definitions, Epilepsia, , March, 1349–1397.