Publikationen Jörg Klepper

Stand 10_2025

1. **Toward a working definition of ketogenic diet resistance in GLUT1 deficiency syndrome**. Falsaperla R, Sortino V, Kluger GJ, Herberhold T, Rüegger A, Striano P, Ruggieri M, Klepper J, Ramantani G. **Epileptic Disord. 2025** Sep 30. doi: 10.1002/epd2.70107. Online ahead of print. PMID: 41026139

2. **Glut1 Deficiency Syndrome: Novel Pathomechanisms, Current Concepts, and Challenges.** Klepper J**. J Inherit Metab Dis. 2025** May;48(3):e70044. doi: 10.1002/jimd.70044. PMID: 40405536 Free PMC article. Review.

3. **Exploring ketogenic diet resistance in glucose transporter type 1 deficiency syndrome: A comprehensive review and critical appraisal.** Falsaperla R, Sortino V, Kluger GJ, Herberhold T, Rüegger A, Striano P, Ruggieri M, Klepper J, Ramantani G. **Epilepsia Open. 2025** Feb;10(1):31-39. doi: 10.1002/epi4.13110. Epub 2024 Dec 6. PMID: 39641282 Free PMC article. Review.

4. **Distinct neurodevelopmental and epileptic phenotypes associated with gain- and loss-of-function GABRB2 variants**. Mohammadi NA, Ahring PK, Yu Liao VW, Chua HC, Ortiz de la Rosa S, Johannesen KM, Michaeli-Yossef Y, Vincent-Devulder A, Meridda C, Bruel AL, Rossi A, Patel C, Klepper J, Bonanni P, Minghetti S, Trivisano M, Specchio N, Amor D, Auvin S, Baer S, Meyer P, Milh M, Salpietro V, Maroofian R, Lemke JR, Weckhuysen S, Christophersen P, Rubboli G, Chebib M, Jensen AA, Absalom NL, Møller RS. **EBioMedicine. 2024** Aug;106:105236. doi: 10.1016/j.ebiom.2024.105236. Epub 2024 Jul 11. PMID: 38996765 Free PMC article.

5. **Diagnosis and treatment of growth hormone deficiency in children on the ketogenic diet: A case series.** Groveman S, Klepper J, Liesenkötter KP, Grimberg A, Bergqvist AGC. **Epilepsia Open. 2024** Jun;9(3):1096-1102. doi: 10.1002/epi4.12942. Epub 2024 Apr 20. PMID: 38642014 Free PMC article.

6. **Exome Sequencing and the Identification of New Genes and Shared Mechanisms in Polymicrogyria.** Akula SK, Chen AY, Neil JE, Shao DD, Mo A, Hylton NK, DiTroia S, Ganesh VS, Smith RS, O'Kane K, Yeh RC, Marciano JH, Kirkham S, Kenny CJ, Song JHT, Al Saffar M, Millan F, Harris DJ, Murphy AV, Klemp KC, Braddock SR, Brand H, Wong I, Talkowski ME, O'Donnell-Luria A, Lai A, Hill RS, Mochida GH, Doan RN, Barkovich AJ, Yang E, Amrom D, Andermann E, Poduri A, Walsh CA; Polymicrogyria Genetics Research Network. **JAMA Neurol. 2023** Sep 1;80(9):980-988. doi: 10.1001/jamaneurol.2023.2363. PMID: 37486637 Free PMC article.

7. **Timing of Ketogenic Dietary Therapy (KDT) Introduction and Its Impact on Cognitive Profiles in Children with Glut1-DS-A Preliminary Study.** Barthold M, Jurkutat A, Goetz R, Schubring L, Spiegler J, Fries AS, Kiesel L, Klepper J. **Children (Basel). 2023** Apr 3;10(4):681. doi: 10.3390/children10040681. PMID: 37189930 Free PMC article.

8. **Treatment of Infantile Spasm Syndrome: Update from the Interdisciplinary Guideline Committee Coordinated by the German-Speaking Society of Neuropediatrics**. Ramantani G, Bölsterli BK, Alber M, Klepper J, Korinthenberg R, Kurlemann G, Tibussek D, Wolff M, Schmitt B. **Neuropediatrics. 2022** Dec;53(6):389-401. doi: 10.1055/a-1909-2977. Epub 2022 Jul 26. PMID: 35882373 Free PMC article.

9. **Consensus statements on the information to deliver after a febrile seizure.** Loussouarn A, Devlin A, Bast T, Benoist G, Corrard F, Cross H, Ferretti A, Viguer FG, Guerrini R, Klepper J, Meissner T, Milh M, Poltorak V, Raucci U, San Antonio-Arce V, Sie A, Smeyers P, Specchio N, Sutcliffe A, Trauffler A, Dozières-Puyravel B, Auvin S. **Eur J Pediatr. 2021** Sep;180(9):2993-2999. doi: 10.1007/s00431-021-04067-2. Epub 2021 Apr 17. PMID: 33866403

10. **Glut1 Deficiency Syndrome (Glut1DS): State of the art in 2020 and recommendations of the international Glut1DS study group.** 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. **Epilepsia Open. 2020** Aug 13;5(3):354-365. doi: 10.1002/epi4.12414. eCollection 2020 Sep. PMID: 32913944 Free PMC article.

11. **Optimal clinical management of children receiving ketogenic parenteral nutrition: a clinical practice guide.** van der Louw E, Aldaz V, Harvey J, Roan M, van den Hurk D, Cross JH, Auvin S; Review Group. **Dev Med Child Neurol. 2020** Jan;62(1):48-56. doi: 10.1111/dmcn.14306. Epub 2019 Jul 16. PMID: 31313290 Free PMC article.

12. **Hemorrhagic Bullous Henoch-Schönlein Purpura: Case Report and Review of the Literature.** Nothhaft M, Klepper J, Kneitz H, Meyer T, Hamm H, Morbach H. **Front Pediatr. 2019** Jan 22;6:413. doi: 10.3389/fped.2018.00413. eCollection 2018. PMID: 30723709 Free PMC article.

13. **SACS variants are a relevant cause of autosomal recessive hereditary motor and sensory neuropathy.** Vill K, Müller-Felber W, Gläser D, Kuhn M, Teusch V, Schreiber H, Weis J, Klepper J, Schirmacher A, Blaschek A, Wiessner M, Strom TM, Dräger B, Hofmeister-Kiltz K, Tacke M, Gerstl L, Young P, Horvath R, Senderek J. **Hum Genet. 2018** Dec;137(11-12):911-919. doi: 10.1007/s00439-018-1952-6. Epub 2018 Nov 21. PMID: 30460542

14. **Benign epilepsy with centrotemporal spikes: Correlating spike frequency and neuropsychology.** Tacke M, Rupp N, Gerstl L, Heinen F, Vill K, Bonfert M, Neubauer BA, Bast T, Borggraefe I; Further Members of the German HEAD Study Group. **Acta Neurol Scand. 2018** Dec;138(6):475-481. doi: 10.1111/ane.13015. Epub 2018 Sep 19. PMID: 30259965

15. **Optimal clinical management of children receiving dietary therapies for epilepsy: Updated recommendations of the International Ketogenic Diet Study Group.** Kossoff EH, Zupec-Kania BA, Auvin S, Ballaban-Gil KR, Christina Bergqvist AG, Blackford R, Buchhalter JR, Caraballo RH, Cross JH, Dahlin MG, Donner EJ, Guzel O, Jehle RS, Klepper J, Kang HC, Lambrechts DA, Liu YMC, Nathan JK, Nordli DR Jr, Pfeifer HH, Rho JM, Scheffer IE, Sharma S, Stafstrom CE, Thiele EA, Turner Z, Vaccarezza MM, van der Louw EJTM, Veggiotti P, Wheless JW, Wirrell EC; Charlie Foundation; Matthew's Friends; Practice Committee of the Child Neurology Society. **Epilepsia Open. 2018** May 21;3(2):175-192. doi: 10.1002/epi4.12225. eCollection 2018 Jun. PMID: 29881797 Free PMC article.

16. **Effects of Levetiracetam and Sulthiame on EEG in benign epilepsy with centrotemporal spikes: A randomized controlled trial**. Tacke M, Borggraefe I, Gerstl L, Heinen F, Vill K, Bonfert M, Bast T; HEAD Study group; Neubauer BA, Baumeister F, Baethmann M, Bentele K, Blank C, Blank HM, Bode H, Bosch F, Brandl U, Brockmann K, Dahlem P, Ernst JP, Feldmann E, Fiedler A, Gerigk M, Heß S, Hikel C, Hoffmann HG, Kieslich M, Klepper J, Kluger G, Koch H, Koch W, Korinthenberg R, Krois I, Kühne H, Kurlemann G, Mandl M, Mause U, Navratil P, Opp J, Penzien J, Prietsch V, Quattländer A, Rating D, Schara U, Shamdeen MG, Sprinz A, Wendker-Magrabi H, Stephani U, Muhle H, Straßburg HM, Töpke B, Trollmann R, Tuschen-Hofstätter E, Waltz S, Weber G, Wien FU, Wolff M, Polster T, Freitag H, Sönmez Ö, Reinhardt K, Traus M, Hoovey Z**. Seizure**. 2018 Mar;56:115-120. doi: 0.1016/j.seizure.2018.01.015. Epub 2018 Feb 3. PMID: 29475094 Free article. Clinical Trial.

17. **Complex care of individuals with multiple sulfatase deficiency: Clinical cases and consensus statement.** Ahrens-Nicklas R, Schlotawa L, Ballabio A, Brunetti-Pierri N, De Castro M, Dierks T, Eichler F, Ficicioglu C, Finglas A, Gaertner J, Kirmse B, Klepper J, Lee M, Olsen A, Parenti G, Vossough A, Vanderver A, Adang LA. **Mol Genet Metab. 2018** Mar;123(3):337-346. doi: 10.1016/j.ymgme.2018.01.005. Epub 2018 Jan 31. PMID: 29397290 Free PMC article.

18. **10 patients, 10 years - Long term follow-up of cardiovascular risk factors in Glut1 deficiency treated with ketogenic diet therapies: A prospective, multicenter case series.** Heussinger N, Della Marina A, Beyerlein A, Leiendecker B, Hermann-Alves S, Dalla Pozza R, Klepper J. **Clin Nutr. 2018** Dec;37(6 Pt A):2246-2251. doi: 10.1016/j.clnu.2017.11.001. Epub 2017 Nov 11. PMID: 29199027 Free article.

19. **Ketogenic Diets in the Treatment of Epilepsy.** Elia M, Klepper J, Leiendecker B, Hartmann H. **Curr Pharm Des. 2017**;23(37):5691-5701. doi: 10.2174/1381612823666170809101517. PMID: 28799513 Review.

20. **Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders.** Wolff M, Johannesen KM, Hedrich UBS, Masnada S, Rubboli G, Gardella E, Lesca G, Ville D, Milh M, Villard L, Afenjar A, Chantot-Bastaraud S, Mignot C, Lardennois C, Nava C, Schwarz N, Gérard M, Perrin L, Doummar D, Auvin S, Miranda MJ, Hempel M, Brilstra E, Knoers N, Verbeek N, van Kempen M, Braun KP, Mancini G, Biskup S, Hörtnagel K, Döcker M, Bast T, Loddenkemper T, Wong-Kisiel L, Baumeister FM, Fazeli W, Striano P, Dilena R, Fontana E, Zara F, Kurlemann G, Klepper J, Thoene JG, Arndt DH, Deconinck N, Schmitt-Mechelke T, Maier O, Muhle H, Wical B, Finetti C, Brückner R, Pietz J, Golla G, Jillella D, Linnet KM, Charles P, Moog U, Õiglane-Shlik E, Mantovani JF, Park K, Deprez M, Lederer D, Mary S, Scalais E, Selim L, Van Coster R, Lagae L, Nikanorova M, Hjalgrim H, Korenke GC, Trivisano M, Specchio N, Ceulemans B, Dorn T, Helbig KL, Hardies K, Stamberger H, de Jonghe P, Weckhuysen S, Lemke JR, Krägeloh-Mann I, Helbig I, Kluger G, Lerche H, Møller RS. **Brain.** 2017 May 1;140(5):1316-1336. doi: 10.1093/brain/awx054. PMID: 28379373

21. **Upstream SLC2A1 translation initiation causes GLUT1 deficiency syndrome.** 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. **Eur J Hum Genet. 2017** Jun;25(6):771-774. doi: 10.1038/ejhg.2017.45. Epub 2017 Apr 5. PMID: 28378819 Free PMC article.

22. **Paroxysmal Nonepileptic Events in Glut1 Deficiency.** Klepper J, Leiendecker B, Eltze C, Heussinger N. **Mov Disord Clin Pract. 2016** Nov-Dec;3(6):607-610. doi: 10.1002/mdc3.12387. Epub 2016 Jul 8. PMID: 28042592 Free PMC article.

23. **Effect of anticonvulsive treatment on neuropsychological performance in children with BECTS.** Tacke M, Gerstl L, Heinen F, Heukaeufer I, Bonfert M, Bast T, Cornell S, Neubauer BA, Borggraefe I; German HEAD Study group**. Eur J Paediatr Neurol. 2016** Nov;20(6):874-879. doi: 10.1016/j.ejpn.2016.07.015. Epub 2016 Jul 25. PMID: 27553576 Clinical Trial.

24. **Ketogenic diet guidelines for infants with refractory epilepsy.** van der Louw E, van den Hurk D, Neal E, Leiendecker B, Fitzsimmon G, Dority L, Thompson L, Marchió M, Dudzińska M, Dressler A, Klepper J, Auvin S, Cross JH. **Eur J Paediatr Neurol. 2016** Nov;20(6):798-809. doi: 10.1016/j.ejpn.2016.07.009. Epub 2016 Jul 17. PMID: 27470655 Free article. Review.

25. **Treatment of Infantile Spasms: Report of the Interdisciplinary Guideline Committee Coordinated by the German-Speaking Society for Neuropediatrics.** Tibussek D, Klepper J, Korinthenberg R, Kurlemann G, Rating D, Wohlrab G, Wolff M, Schmitt B. **Neuropediatrics. 2016** Jun;47(3):139-50. doi: 10.1055/s-0036-1572411. Epub 2016 Feb 24. PMID: 26910805 Review.

26. **Severe Hypertriglyceridemia in Glut1D on Ketogenic Diet.** Klepper J, Leiendecker B, Heussinger N, Lausch E, Bosch F. **Neuropediatrics. 2016** Apr;47(2):132-6. doi: 10.1055/s-0036-1572413. Epub 2016 Feb 22. PMID: 26902182

27. **STXBP1 encephalopathy: A neurodevelopmental disorder including epilepsy.** Stamberger H, Nikanorova M, Willemsen MH, Accorsi P, Angriman M, Baier H, Benkel-Herrenbrueck I, Benoit V, Budetta M, Caliebe A, Cantalupo G, Capovilla G, Casara G, Courage C, Deprez M, Destrée A, Dilena R, Erasmus CE, Fannemel M, Fjær R, Giordano L, Helbig KL, Heyne HO, Klepper J, Kluger GJ, Lederer D, Lodi M, Maier O, Merkenschlager A, Michelberger N, Minetti C, Muhle H, Phalin J, Ramsey K, Romeo A, Schallner J, Schanze I, Shinawi M, Sleegers K, Sterbova K, Syrbe S, Traverso M, Tzschach A, Uldall P, Van Coster R, Verhelst H, Viri M, Winter S, Wolff M, Zenker M, Zoccante L, De Jonghe P, Helbig I, Striano P, Lemke JR, Møller RS, Weckhuysen S. **Neurology. 2016** Mar 8;86(10):954-62. doi: 10.1212/WNL.0000000000002457. Epub 2016 Feb 10. PMID: 26865513 Free article. Review.

28. **GLUT1 deficiency syndrome and ketogenic diet therapies: missing rare but treatable diseases?** Klepper J. **Dev Med Child Neurol. 2015** Oct;57(10):896-7. doi: 10.1111/dmcn.12807. Epub 2015 May 22. PMID: 25997718 Free article. No abstract available.

29. **Chronic Candida albicans Meningitis in a 4-Year-Old Girl with a Homozygous Mutation in the CARD9 Gene (Q295X).** Herbst M, Gazendam R, Reimnitz D, Sawalle-Belohradsky J, Groll A, Schlegel PG, Belohradsky B, Renner E, Klepper J, Grimbacher B, Kuijpers T, Liese J. **Pediatr Infect Dis J. 2015** Sep;34(9):999-1002. doi: 10.1097/INF.0000000000000736. PMID: 25933095

30. **Oligoclonal bands predict multiple sclerosis in children with optic neuritis.** Heussinger N, Kontopantelis E, Gburek-Augustat J, Jenke A, Vollrath G, Korinthenberg R, Hofstetter P, Meyer S, Brecht I, Kornek B, Herkenrath P, Schimmel M, Wenner K, Häusler M, Lutz S, Karenfort M, Blaschek A, Smitka M, Karch S, Piepkorn M, Rostasy K, Lücke T, Weber P, Trollmann R, Klepper J, Häussler M, Hofmann R, Weissert R, Merkenschlager A, Buttmann M; for GRACE-MS (German-speaking Research Alliance for ChildrEn with Multiple Sclerosis). **Ann Neurol. 2015** Jun;77(6):1076-82. doi: 10.1002/ana.24409. Epub 2015 May 11. PMID: 25820181

31. **Exome sequencing identifies a de novo SCN2A mutation in a patient with intractable seizures, severe intellectual disability, optic atrophy, muscular hypotonia, and brain abnormalities.** Baasch AL, Hüning I, Gilissen C, Klepper J, Veltman JA, Gillessen-Kaesbach G, Hoischen A, Lohmann K. **Epilepsia. 2014** Apr;55(4):e25-9. doi: 10.1111/epi.12554. Epub 2014 Mar 1. PMID: 24579881 Review.

32. **Child neurology: differential diagnosis of a low CSF glucose in children and young adults.** Leen WG, de Wit CJ, Wevers RA, van Engelen BG, Kamsteeg EJ, Klepper J, Verbeek MM, Willemsen MA. **Neurology. 2013** Dec 10;81(24):e178-81. doi: 10.1212/01.wnl.0000437294.20817.99. PMID: 24323444 Review.

33. **Mutations in the gene encoding PDGF-B cause brain calcifications in humans and mice**. Keller A, Westenberger A, Sobrido MJ, García-Murias M, Domingo A, Sears RL, Lemos RR, Ordoñez-Ugalde A, Nicolas G, da Cunha JE, Rushing EJ, Hugelshofer M, Wurnig MC, Kaech A, Reimann R, Lohmann K, Dobričić V, Carracedo A, Petrović I, Miyasaki JM, Abakumova I, Mäe MA, Raschperger E, Zatz M, Zschiedrich K, Klepper J, Spiteri E, Prieto JM, Navas I, Preuss M, Dering C, Janković M, Paucar M, Svenningsson P, Saliminejad K, Khorshid HR, Novaković I, Aguzzi A, Boss A, Le Ber I, Defer G, Hannequin D, Kostić VS, Campion D, Geschwind DH, Coppola G, Betsholtz C, Klein C, Oliveira JR. **Nat Genet. 2013** Sep;45(9):1077-82. doi: 10.1038/ng.2723. Epub 2013 Aug 4. PMID: 23913003

34. **Glut1 deficiency syndrome and novel ketogenic diets.** Klepper J, Leiendecker B**. J Child Neurol**. 2013 Aug;28(8):1045-8. doi: 10.1177/0883073813487600. Epub 2013 May 10. PMID: 23666044

35. **Levetiracetam vs. sulthiame in benign epilepsy with centrotemporal spikes in childhood: a double-blinded, randomized, controlled trial (German HEAD Study).** Borggraefe I, Bonfert M, Bast T, Neubauer BA, Schotten KJ, Maßmann K, Noachtar S, Tuxhorn I, May TW, Heinen F; German HEAD Study Group. **Eur J Paediatr Neurol. 2013** Sep;17(5):507-14. doi: 10.1016/j.ejpn.2013.03.014. Epub 2013 Apr 30. PMID: 23642492 Clinical Trial.

36. **Glucide metabolism disorders (excluding glycogen myopathies).** Klepper J. **Handb Clin Neurol. 2013**;113:1689-94. doi: 10.1016/B978-0-444-59565-2.00036-8. PMID: 23622389 Review.

37. **Absence of SLC2A1 mutations does not exclude Glut1 deficiency syndrome.** Klepper J. **Neuropediatrics. 2013** Aug;44(4):235-6. doi: 10.1055/s-0033-1336015. Epub 2013 Mar 12. PMID: 23483445

38. **GLUT-1 deficiency syndrome in Norway--yet another piece of the puzzle.** Klepper J.
39. **Dev Med Child Neurol. 2013** May;55(5):400-1. doi: 10.1111/dmcn.12104. Epub 2013 Mar 1.
40. PMID: 23448598 Free article.

41. **Mutations in SLC20A2 are a major cause of familial idiopathic basal ganglia calcification.** Hsu SC, Sears RL, Lemos RR, Quintáns B, Huang A, Spiteri E, Nevarez L, Mamah C, Zatz M, Pierce KD, Fullerton JM, Adair JC, Berner JE, Bower M, Brodaty H, Carmona O, Dobricić V, Fogel BL, García-Estevez D, Goldman J, Goudreau JL, Hopfer S, Janković M, Jaumà S, Jen JC,

Kirdlarp S, Klepper J, Kostić V, Lang AE, Linglart A, Maisenbacher MK, Manyam BV, Mazzoni P, Miedzybrodzka Z, Mitarnun W, Mitchell PB, Mueller J, Novaković I, Paucar M, Paulson H, Simpson SA, Svenningsson P, Tuite P, Vitek J, Wetchaphanphesat S, Williams C, Yang M, Schofield PR, de Oliveira JR, Sobrido MJ, Geschwind DH, Coppola G. **Neurogenetics. 2013** Feb;14(1):11-22. doi: 10.1007/s10048-012-0349-2. Epub 2013 Jan 20. PMID: 23334463 Free PMC article.

42. **Unusual sensitivity to steroid treatment in intractable childhood epilepsy suggests GLUT1 deficiency syndrome.** Vieker S, Schmitt J, Längler A, Schmidt W, Klepper J. **Neuropediatrics. 2012** Oct;43(5):275-8. doi: 10.1055/s-0032-1324399. Epub 2012 Sep 13. PMID: 22976442

43. **Glucose transporter type 1 deficiency syndrome with carbohydrate-responsive symptoms but without epilepsy.** Koy A, Assmann B, Klepper J, Mayatepek E. **Dev Med Child Neurol. 2011** Dec;53(12):1154-6. doi: 10.1111/j.1469-8749.2011.04082.x. Epub 2011 Aug 12. PMID: 21838819 Free article.

44. **Video/EEG recording of myoclonic absences in GLUT1 deficiency syndrome with a hot-spot R126C mutation in the SLC2A1 gene.** Gökben S, Yılmaz S, Klepper J, Serdaroğlu G, Tekgül H. **Epilepsy Behav. 2011** Jun;21(2):200-2. doi: 10.1016/j.yebeh.2011.03.027. Epub 2011 May 4. PMID: 21546317

45. **GLUT1 deficiency syndrome in clinical practice.** Klepper J. **Epilepsy Res. 2012** Jul;100(3):272-7. doi: 10.1016/j.eplepsyres.2011.02.007. Epub 2011 Mar 5. PMID: 21382692 Review.

46. **Refractory absence epilepsy associated with GLUT-1 deficiency syndrome.** Byrne S, Kearns J, Carolan R, Mc Menamin J, Klepper J, Webb D. **Epilepsia. 2011** May;52(5):1021-4. doi: 10.1111/j.1528-1167.2011.02989.x. Epub 2011 Mar 2. PMID: 21366555

47. **First report of GLUT1 deficiency syndrome in Chinese patients with novel and hot spot mutations in SLC2A1 gene.** Fung EL, Ho YY, Hui J, Wong JH, Ng TB, Fong NY, Klepper J, Tsui KW. **Brain Dev. 2011** Feb;33(2):170-3. doi: 10.1016/j.braindev.2010.03.009. Epub 2010 Apr 22. PMID: 20417043

48. **Anoxic-epileptic seizures.** Stephenson J.B.P. Nechay, A. Klepper J**. J Pediatr Neurol 2010**: 37-38.

49. **Glucose transporter I (GLUT1) Deficiency.** Klepper J. **J Pediatr. Neurol. 2010**: 107-108.

50. **Autosomal recessive inheritance of GLUT1 deficiency syndrome.** Klepper J, Scheffer H, Elsaid MF, Kamsteeg EJ, Leferink M, Ben-Omran T. **Neuropediatrics. 2009** Oct;40(5):207-10. doi: 10.1055/s-0030-1248264. Epub 2010 Mar 10. PMID: 20221955

51. **Glucose transporter-1 deficiency syndrome: the expanding clinical and genetic spectrum of a treatable disorder.** Leen WG, Klepper J, Verbeek MM, Leferink M, Hofste T, van Engelen BG, Wevers RA, Arthur T, Bahi-Buisson N, Ballhausen D, Bekhof J, van Bogaert P, Carrilho I, Chabrol B, Champion MP, Coldwell J, Clayton P, Donner E, Evangeliou A, Ebinger F, Farrell K, Forsyth RJ, de Goede CG, Gross S, Grunewald S, Holthausen H, Jayawant S, Lachlan K, Laugel V, Leppig K, Lim MJ, Mancini G, Marina AD, Martorell L, McMenamin J, Meuwissen ME, Mundy H, Nilsson NO, Panzer A, Poll-The BT, Rauscher C, Rouselle CM, Sandvig I, Scheffner T, Sheridan E, Simpson N, Sykora P, Tomlinson R, Trounce J, Webb D, Weschke B, Scheffer H, Willemsen MA. **Brain. 2010** Mar;133(Pt 3):655-70. doi: 10.1093/brain/awp336. Epub 2010 Feb 2. PMID: 20129935

52. **An electro-encephalogram beta gap after induction with diazepam: a localization method in epileptogenic lesions.** Claus S, Leijten F, Kallansee P, Klepper J, Lopes da Silva FH, Ronner H, Velis D, Viergever MA, Kalitzin S. **Clin Neurophysiol. 2009** Jul;120(7):1235-44. doi: 10.1016/j.clinph.2009.04.007. Epub 2009 May 22. PMID: 19464946

53. **Does delta-sarcoglycan-associated autosomal-dominant cardiomyopathy exist?** Bauer R, Hudson J, Müller HD, Sommer C, Dekomien G, Bourke J, Routledge D, Bushby K, Klepper J, Straub V. **Eur J Hum Genet. 2009** Sep;17(9):1148-53. doi: 10.1038/ejhg.2009.17. Epub 2009 Mar 4. PMID: 19259135 Free PMC article.

54. **Pyridoxal phosphate-dependent neonatal epileptic encephalopathy.** Bagci S, Zschocke J, Hoffmann GF, Bast T, Klepper J, Müller A, Heep A, Bartmann P, Franz AR. **BMJ Case Rep**.**2009**;2009:bcr11.2008.1247. doi: 10.1136/bcr.11.2008.1247. Epub 2009 May 10. PMID: 21686393 Free PMC article.

55. **Glucose transporter deficiency syndrome (GLUT1DS) and the ketogenic diet.** Klepper J. **Epilepsia. 2008** Nov;49 Suppl 8:46-9. doi: 10.1111/j.1528-1167.2008.01833.x. PMID: 19049586 Review.

56. **The ketogenic diet in children with Glut1 deficiency syndrome and epilepsy.** Rauchenzauner M, Klepper J, Leiendecker B, Luef G, Rostasy K, Ebenbichler C. **J Pediatr. 2008** Nov;153(5):716-8. doi: 10.1016/j.jpeds.2008.05.012. PMID: 18940357

57. **Optimal clinical management of children receiving the ketogenic diet: recommendations of the International Ketogenic Diet Study Group.** Kossoff EH, Zupec-Kania BA, Amark PE, Ballaban-Gil KR, Christina Bergqvist AG, Blackford R, Buchhalter JR, Caraballo RH, Helen Cross J, Dahlin MG, Donner EJ, Klepper J, Jehle RS, Kim HD, Christiana Liu YM, Nation J, Nordli DR Jr, Pfeifer HH, Rho JM, Stafstrom CE, Thiele EA, Turner Z, Wirrell EC, Wheless JW, Veggiotti P, Vining EP; Charlie Foundation, Practice Committee of the Child Neurology Society; Practice

Committee of the Child Neurology Society; International Ketogenic Diet Study Group. **Epilepsia. 2009** Feb;50(2):304-17. doi: 10.1111/j.1528-1167.2008.01765.x. Epub 2008 Sep 23. PMID: 18823325

58. **Pyridoxal phosphate-dependent neonatal epileptic encephalopathy.** Bagci S, Zschocke J, Hoffmann GF, Bast T, Klepper J, Müller A, Heep A, Bartmann P, Franz AR. **Arch Dis Child Fetal Neonatal Ed. 2008** Mar;93(2):F151-2. doi: 10.1136/adc.2006.115162. PMID: 18296573

59. **Expect the unexpected: favourable outcome in Munchausen by proxy syndrome.** Klepper J, Heringhaus A, Wurthmann C, Voit T. **Eur J Pediatr. 2008** Sep;167(9):1085-8. doi: 10.1007/s00431-007-0627-4. Epub 2007 Nov 7. PMID: 17987314

60. **Clinical and molecular phenotype of Aicardi-Goutieres syndrome.** Rice G, Patrick T, Parmar R, Taylor CF, Aeby A, Aicardi J, Artuch R, Montalto SA, Bacino CA, Barroso B, Baxter P, Benko WS, Bergmann C, Bertini E, Biancheri R, Blair EM, Blau N, Bonthron DT, Briggs T, Brueton LA, Brunner HG, Burke CJ, Carr IM, Carvalho DR, Chandler KE, Christen HJ, Corry PC, Cowan FM, Cox H, D'Arrigo S, Dean J, De Laet C, De Praeter C, Dery C, Ferrie CD, Flintoff K, Frints SG, Garcia-Cazorla A, Gener B, Goizet C, Goutieres F, Green AJ, Guet A, Hamel BC, Hayward BE, Heiberg A, Hennekam RC, Husson M, Jackson AP, Jayatunga R, Jiang YH, Kant SG, Kao A, King MD, Kingston HM, Klepper J, van der Knaap MS, Kornberg AJ, Kotzot D, Kratzer W, Lacombe D, Lagae L, Landrieu PG, Lanzi G, Leitch A, Lim MJ, Livingston JH, Lourenco CM, Lyall EG, Lynch SA, Lyons MJ, Marom D, McClure JP, McWilliam R, Melancon SB, Mewasingh LD, Moutard ML, Nischal KK, Ostergaard JR, Prendiville J, Rasmussen M, Rogers RC, Roland D, Rosser EM, Rostasy K, Roubertie A, Sanchis A, Schiffmann R, Scholl-Burgi S, Seal S, Shalev SA, Corcoles CS, Sinha GP, Soler D, Spiegel R, Stephenson JB, Tacke U, Tan TY, Till M, Tolmie JL, Tomlin P, Vagnarelli F, Valente EM, Van Coster RN, Van der Aa N, Vanderver A, V… See abstract for full author list ➔ **Am J Hum Genet. 2007** Oct;81(4):713-25. doi: 10.1086/521373. Epub 2007 Sep 4.
61. PMID: 17846997 Free PMC article.

62. **GLUT1 deficiency syndrome--2007 update.** Klepper J, Leiendecker B. **Dev Med Child Neurol. 2007** Sep;49(9):707-16. doi: 10.1111/j.1469-8749.2007.00707.x. PMID: 17718830 Free article. Review.

63. **Exhaled nitric oxide in children after accidental exposure to chlorine gas.** Grasemann H, Tschiedel E, Groch M, Klepper J, Ratjen F. **Inhal Toxicol. 2007** Aug;19(10):895-8. doi: 10.1080/08958370701432157. PMID: 17687720

64. **GLUT1 deficiency with delayed myelination responding to ketogenic diet.** Klepper J, Engelbrecht V, Scheffer H, van der Knaap MS, Fiedler A. **Pediatr Neurol. 2007** Aug;37(2):130-3. doi: 10.1016/j.pediatrneurol.2007.03.009. PMID: 17675029

65. **[The ketogenic diet -- role of the pediatric nurse].** Leiendecker B, Klepper J. **Kinderkrankenschwester. 2006** Dec;25(12):514-7. PMID: 17236684 Review. German. No abstract available.

66. **Mutations in genes encoding ribonuclease H2 subunits cause Aicardi-Goutières syndrome and mimic congenital viral brain infection.** Crow YJ, Leitch A, Hayward BE, Garner A, Parmar R, Griffith E, Ali M, Semple C, Aicardi J, Babul-Hirji R, Baumann C, Baxter P, Bertini E, Chandler KE, Chitayat D, Cau D, Déry C, Fazzi E, Goizet C, King MD, Klepper J, Lacombe D, Lanzi G, Lyall H, Martínez-Frías ML, Mathieu M, McKeown C, Monier A, Oade Y, Quarrell OW, Rittey CD, Rogers RC, Sanchis A, Stephenson JB, Tacke U, Till M, Tolmie JL, Tomlin P, Voit T, Weschke B, Woods CG, Lebon P, Bonthron DT, Ponting CP, Jackson AP. **Nat Genet. 2006** Aug;38(8):910-6. doi: 10.1038/ng1842. Epub 2006 Jul 16. PMID: 16845400

67. **Mutations in the gene encoding the 3'-5' DNA exonuclease TREX1 cause Aicardi-Goutières syndrome at the AGS1 locus.** Crow YJ, Hayward BE, Parmar R, Robins P, Leitch A, Ali M, Black DN, van Bokhoven H, Brunner HG, Hamel BC, Corry PC, Cowan FM, Frints SG, Klepper J, Livingston JH, Lynch SA, Massey RF, Meritet JF, Michaud JL, Ponsot G, Voit T, Lebon P, Bonthron DT, Jackson AP, Barnes DE, Lindahl T. **Nat Genet. 2006** Aug;38(8):917-20. doi: 10.1038/ng1845. Epub 2006 Jul 16. PMID: 16845398

68. **The effects of the ketogenic diet in refractory partial seizures with reference to tuberous sclerosis.** Coppola G, Klepper J, Ammendola E, Fiorillo M, della Corte R, Capano G, Pascotto A. **Eur J Paediatr Neurol. 2006** May;10(3):148-51. doi: 10.1016/j.ejpn.2006.03.001. Epub 2006 Apr 24. PMID: 16632392 Clinical Trial.

69. **Behandelbare seltene metabolische Epilepsien im Kindesalter.** Klepper J, Plecko, B. **Epileptologie 2006**; 23:51-57.

70. Leiendecker B, Klepper J. **Die ketogene Diät – die Role der Kinderkrankenschwester.** Kinderkrankenschwester **2005**, 25. Jg, Nr. 12:514-517.

71. **Seizure control and acceptance of the ketogenic diet in GLUT1 deficiency syndrome: a 2- to 5-year follow-up of 15 children enrolled prospectively.** Klepper J, Scheffer H, Leiendecker B, Gertsen E, Binder S, Leferink M, Hertzberg C, Näke A, Voit T, Willemsen MA. **Neuropediatrics. 2005** Oct;36(5):302-8. doi: 10.1055/s-2005-872843. PMID: 16217704 Clinical Trial.

72. **Bench meets bedside: a 10-year-old girl and amino acid residue glycine 75 of the facilitative glucose transporter GLUT1.** Klepper J, Salas-Burgos A, Gertsen E, Fischbarg J. **Biochemistry. 2005** Sep 27;44(38):12621-6. doi: 10.1021/bi051079t. PMID: 16171377

73. **Clinical presentation, EEG studies, and novel mutations in two cases of GLUT1 deficiency syndrome in Japan.** Ito Y, Gertsen E, Oguni H, Nakayama T, Matsuo M, Funatsuka M, Voit T, Klepper J, Osawa M. **Brain Dev. 2005** Jun;27(4):311-7. doi: 10.1016/j.braindev.2004.09.010. Epub 2004 Dec 9. PMID: 15862198

74. Leiendecker B., Klepper J. **Medikamente und Supplemente bei ketogener Diät im Kindesalter. Neuropädiatrie in Klinik und Praxis, 2005**; 4. Jahrgang Nr.3:98-114.

75. **Genetic heterogeneity in familial idiopathic basal ganglia calcification (Fahr disease).** Oliveira JR, Spiteri E, Sobrido MJ, Hopfer S, Klepper J, Voit T, Gilbert J, Wszolek ZK, Calne DB, Stoessl AJ, Hutton M, Manyam BV, Boller F, Baquero M, Geschwind DH. **Neurology. 2004** Dec 14;63(11):2165-7. doi: 10.1212/01.wnl.0000145601.88274.88. PMID: 15596772 Free article.

76. **[The ketogenic diet in German-speaking countries: update 2003].** Klepper J, Leiendecker B, Riemann E, Baumeister FA. **Klin Padiatr. 2004** Sep-Oct;216(5):277-85. doi: 10.1055/s-2004-44906. PMID: 15455294 German.

77. **Impaired glucose transport into the brain: the expanding spectrum of glucose transporter type 1 deficiency syndrome.** Klepper J. **Curr Opin Neurol. 2004** Apr;17(2):193-6. doi: 10.1097/00019052-200404000-00018. PMID: 15021248 Review.

78. **Effects of the ketogenic diet in the glucose transporter 1 deficiency syndrome.** Klepper J, Diefenbach S, Kohlschütter A, Voit T. **Prostaglandins Leukot Essent Fatty Acids. 2004** Mar;70(3):321-7. doi: 10.1016/j.plefa.2003.07.004. PMID: 14769490

79. Klepper J. **Der Glukosetransporter (GLUT1)-Defekt: Definition einer neuen Erkrankung.** Neuropädiatrie in Klinik und Praxis, **2004**;03:82-86

80. **Die ketogene Diät in den deutschsprachigen Ländern im Jahr 2003: eine Standortbestimmung.** Klepper J, Leiendecker B, Riemann E. Baumeister F.A.M. **Klin Pädiatr 2004**; 216:277-285.

81. **Die ketogene Diät bei Anfallsleiden – Indikationen und Wirkungen.** Klepper J, Leidendecker B. **Aktuel Ernaehr Med 2004**:29:271-274.

82. Functional consequences of an in vivo mutation in exon 10 of the human GLUT1 gene. Lange P, Gertsen E, Monden I, Klepper J, Keller K. **FEBS Lett. 2003** Dec 4;555(2):274-8. doi: 10.1016/s0014-5793(03)01247-x. PMID: 14644427 Free article.

83. **Reversible infantile hypoglycorrhachia: possible transient disturbance in glucose transport?** Klepper J, De Vivo DC, Webb DW, Klinge L, Voit T. **Pediatr Neurol. 2003** Oct;29(4):321-5. doi: 10.1016/s0887-8994(03)00268-6. PMID: 14643395

84. **Mutations in a gene encoding a novel SH3/TPR domain protein cause autosomal recessive Charcot-Marie-Tooth type 4C neuropathy.** Senderek J, Bergmann C, Stendel C, Kirfel J, Verpoorten N, De Jonghe P, Timmerman V, Chrast R, Verheijen MH, Lemke G, Battaloglu E, Parman Y, Erdem S, Tan E, Topaloglu H, Hahn A, Müller-Felber W, Rizzuto N, Fabrizi GM, Stuhrmann M, Rudnik-Schöneborn S, Züchner S, Michael Schröder J, Buchheim E, Straub V, Klepper J, Huehne K, Rautenstrauss B, Büttner R, Nelis E, Zerres K. **Am J Hum Genet. 2003** Nov;73(5):1106-19. doi: 10.1086/379525. Epub 2003 Oct 21. PMID: 14574644 Free PMC article.

85. **Neuroglycopenia in normoglycaemic patients, and the potential benefit of ketosis.** Willemsen MA, Soorani-Lunsing RJ, Pouwels E, Klepper J. **Diabet Med. 2003** Jun;20(6):481-2. doi: 10.1046/j.1464-5491.2003.00952.x. PMID: 12786683

86. **Progressive dystonia in a 12-year-old boy.** Klepper J, Schaper J, Raca G, Coryell J, Das S, Hayflick SJ, Voit T. **Eur J Paediatr Neurol. 2003**;7(2):85-8. doi: 10.1016/s1090-3798(03)00019-9. PMID: 12697433

87. **Introduction of a ketogenic diet in young infants.** Klepper J, Leiendecker B, Bredahl R, Athanassopoulos S, Heinen F, Gertsen E, Flörcken A, Metz A, Voit T. **J Inherit Metab Dis. 2002** Oct;25(6):449-60. doi: 10.1023/a:1021238900470. PMID: 12555938

88. **Effects of anticonvulsants on GLUT1-mediated glucose transport in GLUT1 deficiency syndrome in vitro.** Klepper J, Flörcken A, Fischbarg J, Voit T. **Eur J Pediatr. 2003** Feb;162(2):84-9. doi: 10.1007/s00431-002-1112-8. Epub 2002 Dec 6. PMID: 12548383

89. **Repeated detection of gas in the portal vein after liver transplantation: A sign of EBV-associated post-transplant lymphoproliferation?** Wallot MA, Klepper J, Clapuyt P, Dirsch O, Malagó M, Reding R, Otte JB, Sokal EM. **Pediatr Transplant. 2002** Aug;6(4):332-6. doi: 10.1034/j.1399-3046.2002.02001.x. PMID: 12234275

90. **Facilitated glucose transporter protein type 1 (GLUT1) deficiency syndrome: impaired glucose transport into brain-- a review.** Klepper J, Voit T. **Eur J Pediatr. 2002** Jun;161(6):295-304. doi: 10.1007/s00431-002-0939-3. Epub 2002 Apr 16. PMID: 12029447 Review.

91. **ESPED-Jahresbericht 2001.** Berner R, Bialek, R, Creutzig U, Forster J, Giani G, Hahn A, Klepper J, Nowak-Göttl U, Schaaff F, Schmitt HJ, Schönau E, Tyhen U, Vester U. **DGKJ Mitteilungen 2002** 149 (11):1268-77

92. **Glucose transporter type 1 deficiency syndrome (Glut1DS): methylxanthines potentiate GLUT1 haploinsufficiency in vitro.** Ho YY, Yang H, Klepper J, Fischbarg J, Wang D, De Vivo DC. **Pediatr Res. 2001** Aug;50(2):254-60. doi: 10.1203/00006450-200108000-00015. PMID: 11477212

93. **Functional consequences of the autosomal dominant G272A mutation in the human GLUT1 gene.** Klepper J, Monden I, Guertsen E, Voit T, Willemsen M, Keller K. **FEBS Lett. 2001** Jun 1;498(1):104-9. doi: 10.1016/s0014-5793(01)02463-2. PMID: 11389907 Free article.

94. **Hypoglycorrhachia: a simple clue, simply missed.** Willemsen MA, Verrips A, Verbeek MM, Voit T, Klepper J**. Ann Neurol. 2001** May;49(5):685-6. PMID: 11357965 No abstract available.

95. **Autosomal dominant transmission of GLUT1 deficiency.** Klepper J, Willemsen M, Verrips A, Guertsen E, Herrmann R, Kutzick C, Flörcken A, Voit T. **Hum Mol Genet. 2001** Jan 1;10(1):63-8. doi: 10.1093/hmg/10.1.63. PMID: 11136715

96. **Glucose transporter type 1 deficiency syndrome (glut1ds): methylxanthines potentiate glut1 haploinsufficiency in vitro.** Ho YY, Yang H, Klepper J, Fischbarg J, Wang D, De Vivo DC. **Pediatr Res 2001** 50:254-60

97. **Indikation und Anwendung der ketogenen Diät im Kindesalter** – 1. Workshop ketogene Diät am 15. Dezember 2000 in Essen. Klepper J, Ecker D, Burkart P, Leiendecker B, Voit T, Baumeister FAM. **Monatsschr Kinderheilkd 2001** 149;12:1383-1390.

98. **ESPED-Jahresbericht 2000.** Creutzig U, Forster J, Giani G, Hahn A, Hoffmann GF, Klepper J, von Kries R, Nowak-Göttl U, Schmitt HJ, Thyen U, Wirsing von König CH, Vester U. **Monatsschr Kinderheilkd 2001** 149 (11):1268-77

99. **GLUT1-deficiency: barbiturates potentiate haploinsufficiency in vitro.** Klepper J, Fischbarg J, Vera JC, Wang D, De Vivo DC. **Pediatr Res. 1999** Dec;46(6):677-83. doi: 10.1203/00006450-199912000-00006. PMID: 10590023

100. **Erythrocyte 3-O-methyl-D-glucose uptake assay for diagnosis of glucose-transporter-protein syndrome.** Klepper J, Garcia-Alvarez M, O'Driscoll KR, Parides MK, Wang D, Ho YY, De Vivo DC. **J Clin Lab Anal. 1999**;13(3):116-21. doi: 10.1002/(SICI)1098-2825(1999)13:3<116::AID-JCLA5>3.0.CO;2-C. PMID: 10323476 Free PMC article.

101. **Defective glucose transport across brain tissue barriers: a newly recognized neurological syndrome.** Klepper J, Wang D, Fischbarg J, Vera JC, Jarjour IT, O'Driscoll KR, De Vivo DC. **Neurochem Res. 1999** Apr;24(4):587-94. doi: 10.1023/a:1022544131826. PMID: 10227690

102. **Epilepsy in shunt-treated hydrocephalus**. Klepper J, Büsse M, Strassburg HM, Sörensen N. **Dev Med Child Neurol. 1998** Nov;40(11):731-6. doi: 10.1111/j.1469-8749.1998.tb12340.x. PMID: 9881801 Free article.

103. **Deficient transport of dehydroascorbic acid in the glucose transporter protein syndrome.** Klepper J, Vera JC, De Vivo DC. **Ann Neurol. 1998** Aug;44(2):286-7. doi: 10.1002/ana.410440225. PMID: 9708557 Clinical Trial.

104. **GLUT-1 deficiency syndrome caused by haploinsufficiency of the blood-brain barrier hexose carrier.** Seidner G, Alvarez MG, Yeh JI, O'Driscoll KR, Klepper J, Stump TS, Wang D, Spinner NB, Birnbaum MJ, De Vivo DC. **Nat Genet. 1998** Feb;18(2):188-91. doi: 10.1038/ng0298-188. PMID: 9462754

105. Klepper J, Büsse M, Straßburg H-M, Sörensen N. **Epilepsy in shunt-treated hydrocephalus. Develop Med Child Neurol 1998** 11:731-736.

106. **Technical note: Rationale, development, use and evaluation of an equipment management and image storage system.** Klepper J, Geiger G. **J Clin Eng. 1996** Jul-Aug;21(4):313-6. doi: 10.1097/00004669-199607000-00016. PMID: 10159653

107. **Antizipation und Trinukleotidexpansion bei juveniler Chorea Huntington.** Klepper J, Spiegel R, Straßburg H-M, Huppertz HI. **Monatsschr Kinderheilkd 1994** 142:127-129.

108. **Biochemical assessment and clinical evaluation of a bilirubin adsorbent column (BR-350) in critically ill patients with intractable jaundice.** Geiger H, Klepper J, Lux P, Heidland A. **Int J Artif Organs. 1992** Jan;15(1):35-9. PMID: 1551726

Mitarbeit an AWMF-Leitlinien:

- AWMF-Leitlinie **„Ketogene Ernährungstherapien (KET)“** – federführender Autor.
  S1 Leitlinie der Gesellschaft für Neuropädiatrie, Version: 4.1, Stand: 30.11.**2021**
  AWMF-Leitlinien-Register Nr. 022/021 Entwicklungsstufe: 1
  Gültig bis: 29.11.2026

- AWMF-Leitlinie „**Therapie der Blitz-Nick-Salaam Epilepsie (West-Syndrom)“** – Coautor.
  S3 Leitlinie der Gesellschaft für Neuropädiatrie, Version: 3.0, Aktualisierung Februar **2021**
  AWMF-Register Nr. 022/022 Klasse: S3
  Gültig bis: 27.02.2026

- AWMF-Leitlinie **„Diagnostisches Vorgehen bei Verdacht auf neurodegenerative oder neurometabolische Krankheit**. – Coautor. S1 Leitlinie der Gesellschaft für Neuropädiatrie,
  AWMF-Leitlinien-Register Nr. 022/018 Entwicklungsstufe: 1
  Gültig bis: abgelaufen **(2010)**

Buchbeiträge:

Klepper, J., Kapitel E 13, S. 410-413. **Ketogene Ernährungstherapie**. In: Gerstl/Borggräfe/Hufschmidt/Heinen (Hrsg.). Pädiatrische Neurologie – Neuropädiatrie. Praxishandbuch und Atlas. 2., erweiterte und überarbeitete Auflage. **2025**. ISBN 978-3-17-036226-0, 1362 Seiten mit 228 Abb., 185 Tab., Verlag W. Kohlhammer.

Klepper J. Kapitel 12, S 282-286. **Anwendung ketogener Ernährungstherapien in der Neuropädiatrie.** In: Angela M. Kaindl (ed.), Neuropädiatrie. Praxishandbuch. ISBN 978-3-8374-1655-8 928 Seiten Deutsch / 6. Auflage **2023**, UNI-MED (Verlag).

Klepper J. Section I, Chapter 6, p.43-50. **Glut1 Deficiency Syndrome and the ketogenic dietary therapies.** In: Susan A. Masino (ed.). Ketogenic Diet and Metabolic Therapies. Expanded Roles in Health and Disease. ISBN-13: 978-0197501207, 566 Seiten Englisch / 2nd Edition **2022**, Oxford University Press.

Klepper J. Kap. 42.2., S. 824-833. **Anwendung ketogener Diäten in der Neuropädiatrie.** In: Fuat Aksu (ed). Neuropädiatrie. Praxishandbuch. ISBN: 978-3-8374-1578-0 896 Seiten Deutsch / 5. Auflage **2019**, UNI-MED (Verlag).

Klepper J, Leiendecker B., Section 1, Chapter 5, pp. 35-41. **Disorders of carbohydrate metabolism. GLUT1 Deficiency**. In: Hollak C.E.M, Lachmann R.H. (eds). Inherited metabolic disease in adults. A clinical guide. ISBN: 9780199972135 **2017**, Oxford University Press.

Klepper J. Section I, Chapter 5, p.35-40. **Glut1 Deficiency and the ketogenic diets**. In: Susan A. Masino (ed.). Ketogenic Diet and Metabolic Therapies. Expanded Roles in Health and Disease. ISBN-10 : 9780190497996, 424 Seiten Englisch / 1st Edition **2017**, Oxford University Press.

Santer R, Klepper J. Section II, Chapter 10, p. 175-182. **Disorders of Glucose Transport.** In: J.-M. Saudubray, M.R. Baumgartner, J. Walter (Eds.). Inborn Metabolic Diseases. Diagnosis and Treatment. ISBN 978-3-662-49769-2, 7th edition, 658 Seiten Englisch / Erscheinungsdatum: 28.10.**2016,** Springer Verlag.

Klepper J. **Glucide metabolism disorders (exlcuding gylcogen myopathies).** Chapter 171, p. 1689-1695. In: Pediatric Neurology, 187 Chapters, O. Dulac, H. Sarnat, M. Lasonde (Eds). A volume in: Handbook of Clinical Neurology, ISBN 9780444529107,1937 Seiten Englisch, 3rd Series **2013**, Elsevier, Edinburgh.

Santer R, Klepper J. Section II, Chapter 11, p. 175-181. **Disorders of Glucose Transport**. In: J.-M. Saudubray, M.R. Baumgartner, J. Walter (Eds.). Inborn Metabolic Diseases. Diagnosis and Treatment. ISBN-13 978-3-642-15719-6, 5th edition, 656 Seiten, Englisch / Erscheinungsdatum: **2012**, Springer Verlag.

Klepper J. Leiendecker B. Kossoff E.H. **Pocket guide to the ketogenic diet.** 2nd edition **2007**, ISBN 978-3-936145-51-9

Santer R, Klepper J. Chapter 16. **Disorders of Glucose Transport.** In: N. Blau, G.F. Hoffmann, J. Leonard, J.T.R. Clarke (Eds.). Physician's Guide to the Treatment and Follow-Up of Metabolic Diseases. ISBN-13 978-3-540-22954-4. 415 SAeiten, Englisch / Erscheinungsdatum **2006**, Springer-Verlag.

Klepper J (ed.). **Brain metabolism revisited - concepts and treatments – metabolic epileptic encephalopathies.** SPS Verlagsgesellschaft Heilbronn, **2005,** ISBN 3-936145-34-2.

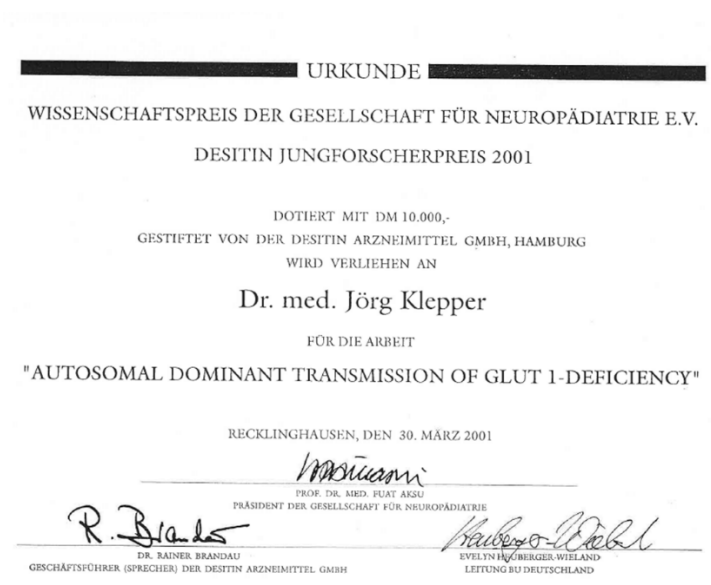
Santer R, Klepper J. Chapter 11. **Disorders of Glucose Transport.** In: F.J. Saudubray, J.M. G. van den Berghe (Eds.). Inborn Metabolic Diseases – diagnosis and treatment. 4th edition**, 2005**.

Auszeichnungen:

Wissenschaftspreis der Gesellschaft für Neuropädiatrie e.V., 2001: Dr. med. Jörg Klepper: **„Autosomal dominant transmission of GLUT1-deficiency“.**

Ziel des Wissenschaftspreises ist die Förderung von Forschungsprojekten der Jungforscher auf dem Gebiet Neuropädiatrie, die einen unmittelbaren Bezug zu klinischen Problemen aufweisen.

John Freeman-Award 2025: Prof. Dr. med. Jörg Klepper, im September 2025 auf dem 9. Symposium für ketogene Therapien, Paris. „*For exemplary dedication to advancing the clinical use and scientific understanding of Medical Ketogenic Dietary Therapies*.“



URKUNDE

WISSENSCHAFTSPREIS DER GESELLSCHAFT FÜR NEUROPÄDIATRIE E.V.

DESITIN JUNGFORSCHERPREIS 2001

DOTIERT MIT DM 10.000,-
GESTIFTET VON DER DESITIN ARZNEIMITTEL GMBH, HAMBURG
WIRD VERLIEHEN AN

Dr. med. Jörg Klepper

FÜR DIE ARBEIT

"AUTOSOMAL DOMINANT TRANSMISSION OF GLUT 1-DEFICIENCY"

RECKLINGHAUSEN, DEN 30. MÄRZ 2001

PROF. DR. MED. FUAT AKSU
PRÄSIDENT DER GESELLSCHAFT FÜR NEUROPÄDIATRIE

DR. RAINER BRANDAU
GESCHÄFTSFÜHRER (SPRECHER) DER DESITIN ARZNEIMITTEL GMBH

EVELYN HAUBERGER-WIELAND
LEITUNG BU DEUTSCHLAND