Ausgewählte Publikationen 2012-2018

2018

Schatz UA, Weiss S, Wenninger S, Schoser B, Muss WH, Bittner RE, Schmidt WM, Schossig AS, Rudnik-Schöneborn S, Baumann M. **Evidence of mild founder LMOD3 mutations causing nemaline myopathy 10 in Germany and Austria**. Neurology. 2018 Oct 5. doi: 10.1212/WNL.0000000000006428. [Epub ahead of print] PubMed PMID: 30291184.

Baumann M, Schreiber H, Schlotter-Weigel B, Löscher WN, Stucka R, Karall D, Strom TM, Bauer P, Krabichler B, Fauth C, Glaeser D, Senderek J. **MPV17 mutations in juvenile- and adult-onset axonal sensorimotor polyneuropathy**. Clin Genet. 2018 Oct 9. doi: 10.1111/cge.13462. [Epub ahead of print] PubMed PMID: 30298599.

Ruzsányi V, Péter Kalapos M, Schmidl C, Karall D, Scholl-Bürgi S, Baumann M (2018): **Breath profiles of children on ketogenic therapy**. J Breath Res 12: 036021.

Baumann M, Grams A, Djurdjevic T, Hennes EM, Lechner C, Behring B, Blaschek A, Diepold K, Eisenkölbl A, Fluss J, Karenfort M, Koch J, Konuskan B, Leiz S, Merkenschlager A, Pohl D, Schimmel M, Thiels C, Kornek B, Schanda K, Reindl M, Rostásy K (2018): **MRI of the first event in pediatric acquired demyelinating syndromes with antibodies to myelin oligodendrocyte glycoprotein**; J Neurol 265: 845-855.

Hacohen Y, Wong YY, Lechner C, Jurynczyk M, Wright S, Konuskan B, Kalser J, Poulat AL, Maurey H, Ganelin-Cohen E, Wassmer E, Hemingway C, Forsyth R, Hennes EM, Leite MI, Ciccarelli O, Anlar B, Hintzen R, Marignier R, Palace J, Baumann M, Rostásy K, Neuteboom R, Deiva K, Lim M (2018): **Disease course and treatment responses in children with relapsing myelin oligodendrocyte glycoprotein antibody-associated disease**; JAMA Neurol 265: 845-855.

2017

Hennes EM, Baumann M, Schanda K, Anlar B, Bajer-Kornek B, Blaschek A, Brantner-Inthaler S, Diepold K, Eisenkölbl A, Gotwald T, Kuchukhidze G, Gruber-Sedlmayr U, Häusler M, Höftberger R, Karenfort M, Klein A, Koch J, Kraus V, Lechner C, Leiz S, Leypoldt F, Mader S, Marquard K, Poggenburg I, Pohl D, Pritsch M, Raucherzauner M, Schimmel M, Thiels C, Tibussek D, Vieker S, Zeches C, Berger T, Reindl M, Rostásy K; BIOMARKER Study Group (2017): **Prognostic relevance of MOG antibodies in children with an acquired demyelinating syndrome**; Neurology 89: 900-908.

Giunta C, Baumann M, Fauth C, Lindert U, Abdalla EM, Brady AF, Collins J, Dastgir J, Donkervoort S, Ghali N, Johnson DS, Kariminejad A, Koch J, Kraenzlin M, Lahiri N, Lozic B, Manzur AY, Morton JEV, Pilch J, Pollitt RC, Schreiber G, Shannon NL, Sobey G, Vandersteen A, van Dijk FS, Witsch-Baumgartner M, Zschocke J, Pope FM, Bönnemann CG, Rohrbach M (2017): **A cohort of 17 patients with kyphoscoliotic Ehlers-Danlos syndrome caused by biallelic mutations in FKBP14: expansion of the clinical and mutational spectrum and description of the natural history**; Genet Med 20: 42-54.

Baumann M, Steichen-Gersdorf E, Krabichler B, Müller T, Janecke AR (2017): **A recognizable type of syndromic short stature with arthrogryposis caused by bi-allelic SEMA3A loss-of-function variants**; Clin Genet 92: 86-90.

Baumann M, Steichen-Gersdorf E, Krabichler B, Petersen BS, Weber U, Schmidt WM, Zschocke J, Müller T, Bittner RE, Janecke AR (2017): **Homozygous SYNE1 mutation causes congenital onset of muscular weakness with distal arthrogryposis: a genotype-phenotype correlation**; Eur J Hum Genet 25: 262-266.

2016

Rauchenzauner M, Hagn C, Walch R, Baumann M, Haberlandt E, Frühwirth M, Rostasy K (2017): **Quality of Life and Fitness in Children and Adolescents with Epilepsy (EpiFit)**; Neuropediatrics 48: 161-165.

Baumann M, Hennes EM, Schanda K, Karenfort M, Kornek B, Seidl R, Diepold K, Lauffer H, Marquardt I, Strautmanis J, Syrbe S, Vieker S, Höftberger R, Reindl M, Rostásy K. **Children with multiphasic disseminated encephalomyelitis and antibodies to the myelin oligodendrocyte glycoprotein (MOG): Extending the spectrum of MOG antibody positive diseases.** Mult Scler. 2016 Dec;22(14):1821-1829. PubMed PMID: 26869530.

Lechner C, Baumann M, Hennes EM, Schanda K, Marquard K, Karenfort M, Leiz S, Pohl D, Venkateswaran S, Pritsch M, Koch J, Schimmel M, Häusler M, Klein A, Blaschek A, Thiels C, Lücke T, Gruber-Sedlmayr U, Kornek B, Hahn A, Leypoldt F, Sandrieser T, Gallwitz H, Stoffels J, Korenke C, Reindl M, Rostásy K. **Antibodies to MOG and AQP4 in children with neuromyelitis optica and limited forms of the disease.** J Neurol Neurosurg Psychiatry. 2016 Aug;87(8):897-905. doi: 10.1136/jnnp-2015-311743. Epub 2015 Dec 8. PubMed PMID: 26645082.

Sellier E, Platt MJ, Andersen GL, Krägeloh-Mann I, De La Cruz J, Cans C; Surveillance of Cerebral Palsy Network. **Decreasing prevalence in cerebral palsy: a multi-site European population-based study, 1980 to 2003.** Dev Med Child Neurol. 2016 Jan;58(1):85-92. doi: 10.1111/dmcn.12865. Epub 2015 Aug 28. PubMed PMID: 26330098.

Zlamy M, Haberlandt E, Brunner J, Dozcy L, Rostasy K. **Tick-borne encephalitis in a child with previous history of completed primary vaccination.** Pediatr Int. 2016 Jan;58(1):56-8. doi: 10.1111/ped.12752. Epub 2015 Nov 6. PubMed PMID: 26541246.

2015

Baumann M, Sahin K, Lechner C, Hennes EM, Schanda K, Mader S, Karenfort M, Selch C, Häusler M, Eisenkölbl A, Salandin M, Gruber-Sedlmayr U, Blaschek A, Kraus V, Leiz S, Finsterwalder J, Gotwald T, Kuchukhidze G, Berger T, Reindl M, Rostásy K. **Clinical and neuroradiological differences of paediatric acute disseminating encephalomyelitis with and without antibodies to the myelin oligodendrocyte glycoprotein.** J Neurol Neurosurg Psychiatry. 2015 Mar;86(3):265-72. doi: 10.1136/jnnp-2014-308346. Epub 2014 Aug 13. PubMed PMID: 25121570.

Michel M, Haberlandt E, Baumann M, Entenmann A, Wagner M, Rostasy K. **Postdural Puncture Superior Sagittal Sinus Thrombosis in a Juvenile Case of Clinically Isolated Syndrome.** Case Rep Pediatr. 2015;2015:358164. doi: 10.1155/2015/358164. Epub 2015 Oct 19. PubMed PMID: 26558127; PubMed Central PMCID: PMC4629016. *Free full text*

Ramberger M, Bsteh G, Schanda K, Höftberger R, Rostásy K, Baumann M, Aboulenein-Djamshidian F, Lutterotti A, Deisenhammer F, Berger T, Reindl M. **NMDA receptor antibodies: A rare association in**

inflammatory demyelinating diseases. Neurol Neuroimmunol Neuroinflamm. 2015 Aug 13;2(5):e141. doi: 10.1212/NXI.0000000000000141. eCollection 2015 Oct. PubMed PMID: 26309901; PubMed Central PMCID: PMC4537309. *Free full text*

Scholl-Bürgi S, Höller A, Pichler K, Michel M, Haberlandt E, Karall D. **Ketogenic diets in patients with inherited metabolic disorders.** J Inherit Metab Dis. 2015 Jul;38(4):765-73. doi: 10.1007/s10545-015-9872-2. Epub 2015 Jun 25. Review. PubMed PMID: 26109259.

2014

Haberlandt E, Karall D, Jud V, Baumgartner SS, Zotter S, Rostasy K, Baumann M, Scholl-Buergi S. **Glucose transporter type 1 deficiency syndrome effectively treated with modified Atkins diet.** Neuropediatrics. 2014 Apr;45(2):117-9. doi: 10.1055/s-0033-1349225. Epub 2013 Jul 25. PubMed PMID: 23888468.

Karall D, Scholl-Bürgi S, Widmann G, Albrecht U, Niedermayr K, Maurer K, Ausserer B, Huemer M, Bale R. **Stereotactic radiofrequency ablation for liver tumors in inherited metabolic disorders.** Cardiovasc Intervent Radiol. 2014 Aug;37(4):1027-33. doi: 10.1007/s00270-013-0756-2. Epub 2013 Oct 23. PubMed PMID: 24149831.

Rauchenzauner M, Roscia S, Prieschl M, Wildt L, Haberlandt E, Baumann M, Rostasy K, Agostinelli S, Pizzolorusso A, Luef G, Verrotti A. **Reproductive endocrine health in pubertal females with epilepsy on antiepileptic drugs: time to screen?** Neuropediatrics. 2014 Aug;45(4):226-33. doi: 10.1055/s-0033-1364104. Epub 2014 Jan 9. PubMed PMID: 24407471.

Spreiz A, Haberlandt E, Baumann M, Baumgartner Sigl S, Fauth C, Gautsch K, Karall D, Janetschek C, Rostasy K, Scholl-Bürgi S, Zotter S, Utermann G, Zschocke J, Kotzot D. **Chromosomal microaberrations in patients with epilepsy, intellectual disability, and congenital anomalies.** Clin Genet. 2014 Oct;86(4):361-6. doi: 10.1111/cge.12288. Epub 2013 Oct 28. PubMed PMID: 24116836.

2013

Finsterer J, Löscher W, Wanschitz J, Baumann M, Quasthoff S, Grisold W. **Hirayama disease in Austria.** Joint Bone Spine. 2013 Oct;80(5):503-7. doi: 10.1016/j.jbspin.2012.10.013. Epub 2012 Dec 10. PubMed PMID: 23237996.

Haberlandt E, Rauchenzauner M, Morass M, Wondrak P, Scholl-Bürgi S, Rostásy K, Karall D. **Matrix-metalloproteinases and proinflammatory cytokines in children with febrile convulsions and epilepsy - cause or consequence?** Epilepsy Res. 2013 Jul;105(1-2):225-8. doi: 10.1016/j.eplepsyres.2012.12.003. Epub 2013 Jan 10. PubMed PMID: 23313321.

Kiechl-Kohlendorfer U, Ralser E, Pupp Peglow U, Pehboeck-Walser N, Fussenegger B. **Early risk predictors for impaired numerical skills in 5-year-old children born before 32 weeks of gestation.** Acta Paediatr. 2013 Jan;102(1):66-71. doi: 10.1111/apa.12036. Epub 2012 Oct 22. PubMed PMID: 23009635.

Krupp LB, Tardieu M, Amato MP, Banwell B, Chitnis T, Dale RC, Ghezzi A, Hintzen R, Kornberg A, Pohl D, Rostasy K, Tenembaum S, Wassmer E; **International Pediatric Multiple Sclerosis Study Group. International Pediatric Multiple Sclerosis Study Group criteria for pediatric multiple sclerosis and immune-mediated central nervous system demyelinating disorders: revisions to the 2007**

definitions. Mult Scler. 2013 Sep;19(10):1261-7. doi: 10.1177/1352458513484547. Epub 2013 Apr 9. PubMed PMID: 23572237.

Müller-Felber W, Wanschitz J, Vill K, Baumann M. **Pediatric idiopathic inflammatory myopathies: an update on diagnostic and treatment strategies.** Neuropediatrics. 2013 Dec;44(6):314-23. doi: 10.1055/s-0033-1358600. Epub 2013 Nov 7. Review. PubMed PMID: 24203857.

Rostásy K, Mader S, Hennes EM, Schanda K, Gredler V, Guenther A, Blaschek A, Korenke C, Pritsch M, Pohl D, Maier O, Kuchukhidze G, Brunner-Krainz M, Berger T, Reindl M. **Persisting myelin oligodendrocyte glycoprotein antibodies in aquaporin-4 antibody negative pediatric neuromyelitis optica.** Mult Scler. 2013 Jul;19(8):1052-9. doi: 10.1177/1352458512470310. Epub 2012 Dec 20. PubMed PMID: 23257621.

Weckhuysen S, Ivanovic V, Hendrickx R, Van Coster R, Hjalgrim H, Møller RS, Grønborg S, Schoonjans AS, Ceulemans B, Heavin SB, Eltze C, Horvath R, Casara G, Pisano T, Giordano L, Rostasy K, Haberlandt E, Albrecht B, Bevot A, Benkel I, Syrbe S, Sheidley B, Guerrini R, Poduri A, Lemke JR, Mandelstam S, Scheffer I, Angriman M, Striano P, Marini C, Suls A, De Jonghe P; KCNQ2 Study Group. **Extending the KCNQ2 encephalopathy spectrum: clinical and neuroimaging findings in 17 patients.** Neurology. 2013 Nov 5;81(19):1697-703. doi: 10.1212/01.wnl.0000435296.72400.a1. Epub 2013 Oct 9. PubMed PMID: 24107868; PubMed Central PMCID: PMC3812107. *Free full text*

Zotter S, Koch J, Schlachter K, Katzensteiner S, Dorninger L, Brunner J, Baumann M, Wolf-Magele A, Schmid H, Ulmer H, Hagspiel S, Rostasy K. **Neuropsychological profile of children after an episode of neuroborreliosis.** Neuropediatrics. 2013 Dec;44(6):346-53. doi: 10.1055/s-0033-1349724. Epub 2013 Aug 6. PubMed PMID: 23921969.

2012

Baumann M, Giunta C, Krabichler B, Rüschendorf F, Zoppi N, Colombi M, Bittner RE, Quijano-Roy S, Muntoni F, Cirak S, Schreiber G, Zou Y, Hu Y, Romero NB, Carlier RY, Amberger A, Deutschmann A, Straub V, Rohrbach M, Steinmann B, Rostásy K, Karall D, Bönnemann CG, Zschocke J, Fauth C. **Mutations in FKBP14 cause a variant of Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss.** Am J Hum Genet. 2012 Feb 10;90(2):201-16. doi: 10.1016/j.ajhg.2011.12.004. Epub 2012 Jan 19. PubMed PMID: 22265013; PubMed Central PMCID: PMC3276673. *Free full text*

Hennes E, Zotter S, Dorninger L, Hartmann H, Häusler M, Huppke P, Jacobs J, Kraus V, Makowski C, Schlachter K, Ulmer H, van Baalen A, Koch J, Gotwald T, Rostasy K. **Long-term outcome of children with acute cerebellitis.** Neuropediatrics. 2012 Oct;43(5):240-8. doi: 10.1055/s-0032-1324732. Epub 2012 Aug 30. PubMed PMID: 22936351.

Karall D, Haberlandt E, Albrecht U, Rostasy K, Häberle J, Scholl-Bürgi S. **Unrecognized citrullinemia mimicking encephalitis in a 14-year-old boy: unexpected result through the use of a standardized lumbar puncture protocol.** Neuropediatrics. 2012 Apr;43(2):59-63. doi: 10.1055/s-0032-1307453. Epub 2012 Apr 3. PubMed PMID: 22473243.

King J, Mochalski P, Unterkofler K, Teschl G, Klieber M, Stein M, Amann A, Baumann M. **Breath isoprene: muscle dystrophy patients support the concept of a pool of isoprene in the periphery of the human body.** Biochem Biophys Res Commun. 2012 Jul 6;423(3):526-30. doi:

10.1016/j.bbrc.2012.05.159. Epub 2012 Jun 5. PubMed PMID: 22683640; PubMed Central PMCID: PMC4959562. *Free full text*

Krabichler B, Rostasy K, Baumann M, Karall D, Scholl-Bürgi S, Schwarzer C, Gautsch K, Spreiz A, Kotzot D, Zschocke J, Fauth C, Haberlandt E. **Novel mutation in potassium channel related gene KCTD7 and progressive myoclonic epilepsy.** Ann Hum Genet. 2012 Jul;76(4):326-31. doi: 10.1111/j.1469-1809.2012.00710.x. Epub 2012 May 21. PubMed PMID: 22606975. *Free full text*

Rostasy K, Mader S, Schanda K, Huppke P, Gärtner J, Kraus V, Karenfort M, Tibussek D, Blaschek A, Bajer-Kornek B, Leitz S, Schimmel M, Di Pauli F, Berger T, Reindl M. **Anti-myelin oligodendrocyte glycoprotein antibodies in pediatric patients with optic neuritis.** Arch Neurol. 2012 Jun;69(6):752-6. doi: 10.1001/archneurol.2011.2956. PubMed PMID: 22371853.

Schossig A, Wolf NI, Fischer C, Fischer M, Stocker G, Pabinger S, Dander A, Steiner B, Tönz O, Kotzot D, Haberlandt E, Amberger A, Burwinkel B, Wimmer K, Fauth C, Grond-Ginsbach C, Koch MJ, Deichmann A, von Kalle C, Bartram CR, Kohlschütter A, Trajanoski Z, Zschocke J. **Mutations in ROGDI Cause Kohlschütter-Tönz Syndrome.** Am J Hum Genet. 2012 Apr 6;90(4):701-7. doi: 10.1016/j.ajhg.2012.02.012. Epub 2012 Mar 15. PubMed PMID: 22424600; PubMed Central PMCID: PMC3322220. *Free full text*