Publikationen durch Mitarbeiter des Sozialpädiatrischen Zentrums Neuropädiatrie in den letzten 5 Jahren

2015

Originalartikel

Baxter PS, Bell KF, Hasel P, Kaindl AM, Fricker M, Thomson D, Cregan SP, Gillingwater TH, Hardingham GE. Synaptic NMDA receptor activity is coupled to the transcriptional control of the glutathione system in the developing brain. *Nat Commun* 2015;6:6761. **IF 11,470**

Schottmann G, Jungbluth H, Schara U, Knierim E, Morales Gonzalez S, Gill E, Seifert F, Norwood F, Deshpande C, von Au K, Schuelke M, Senderek J. Recessive truncating IGHMBP2 mutations presenting as axonal sensorimotor neuropathy. *Neurology* 2015;84(5):523-31. **IF 8,303**

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Adegbola A, Musante L, Callewaert B, Maciel P, Hu H, Isidor B, Minh-Picker S, Le Caignec C, Delle Chiaie B, Vanakker O, Menten B, D'Heedene A, Bockaert N, Roelens F, Decaestecker K, Silva J, Soares G, Lopes F, Najmabadi H, Kahrizi K, Cox GF, Angus SP, Fischer U, Suckow V, Bartsch O, Chess A, Ropers HH, Wienker TF, Hübner Ch, Kaindl AM, Kalscheuer VM. Redefining the MED13L syndrome. *EJHG* 2015. Im Druck. **IF 4,349**

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Ottenhausen M, Bittigau P, Koch A, Lankes E, Schulz M, Thomale UW. Hamartoma of the optic nerve in a young child-case report and review of the literature. *Childs Nerv Syst* 2015;31(8):1401-6. **IF 1,163**

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Morris-Rosendahl D, Kaindl AM. Generation Sequencing (NGS) technology has enabled us to learn about primary autosomal recessive microcephaly (MCPH). *Mol Cell Probes.* 2015. Im Druck. **IF 1,859**

Buchkapitel

Passemard S, Verloes A, Kaindl AM. Microcephaly. In: Dulac O, Sarnat H, Lassonde M (Eds.). Handbook of Clinical Neurology: Pediatric Neurology. Elsevier, Masson, Frankreich.

Hübner C, Kaindl AM, Schülke M. Neuromuskuläre Erkrankungen. In: Speer CP, Gahr M (Eds.). Pädiatrie, 4. Auflage, Springer Medizinverlag, Heidelberg, Germany.

Schwabe G, Baechli H, Boltshauser E, Kaindl AM. Entwicklungsstörungen des Nervensystems. In: Hoffmann G (Ed.). Facharztbuch Pädiatrie. Springer Medizinverlag, Heidelberg, Germany.

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2014

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Cottenie E, Kochanski A, Jordanova A, Bansagi B, Zimon M, Horga A, Jaunmuktane Z, Saveri P, Rasic VM, Baets J, Bartsakoulia M, Ploski R, Teterycz P, Nikolic M, Quinlivan R, Laura M, Sweeney MG, Taroni F, Lunn MP, Moroni I, Gonzalez M, Hanna MG, Bettencourt C, Chabrol E, Franke A, von Au K, Schilhabel M, Kabzińska D, Hausmanowa-Petrusewicz I, Brandner S, Lim SC, Song H, Choi BO, Horvath R, Chung KW, Zuchner S, Pareyson D, Harms M, Reilly MM, Houlden H. Truncating and missense mutations in *IGHMBP2* cause Charcot-Marie Tooth disease type 2. *Am J Hum Genet* 2014;95(5):590-601. **IF 10.987**

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Gonnermann J, Klamann MK, Maier AK, Bertelmann E, Schroeter J, von Au K, Joussen AM, Torun N. Descemet membrane endothelial keratoplasty in a child with corneal endothelial dysfunction in Kearns-Sayre syndrome. *Cornea* 2014;33(11):1232-4. **IF 2.36**

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Lohkamp LN, von Au K, Goebel HH, Kress W, Grieben U, Drossel K, Garbes L, Wirth B, Heppner FL, Stenzel W. A paucisymptomatic neuromuscular disease mimicking type III 5q-SMA with complex rearrangements in the SMN gene. *J Child Neurol* 2014;29(2):254-9. **IF 1.717**

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2013

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Degos V, Peineau S, Nijboer C, Kaindl AM, Sigaut S, Favrais G, Plaisant F, Tessier N, Gouadon E, Lombet A, Saliba E, Collingridge GL, Maze M, Nicoletti F, Heijnen C, Mantz J, Kavelaars A, Gressens P. A novel mechanism underlying inflammation-induced excitotoxic neurodegenration. *Ann Neurol* 2013;73(5):667-78. **IF 11.91**

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Ohlraun S, Wollersheim T, Weiß C, Markus P, Weber-Carstens S, Schmitz D, Schuelke M. *CARbon DIoxide for the treatment of Febrile seizures: rationale, feasibility, and design of the CARDIF-study. J Transl Med* 2013;11:157. **IF 3,991**

Issa L, Mueller K, Seufert K, Kraemer N, Rosenkotter H, Ninnemann O, Buob M, Kaindl AM* Morris-Rosendahl D.* Clinical and cellular features in patients with primary autosomal recessive microcephaly and a novel *CDK5RAP2* mutation. *Orphanet J Rare Dis* 2013;8:59. **IF 3.958**

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Kaindl AM, Degos V, Peineau S, Gouadon E, Loron G, Collingridge GL, Lombet A, Kavelaars A, Verney C, Mantz J, Gressens P. NMDA receptors are expressed in microglia, regulate the inflammatory response and mediate neuronal injury. *Ann Neurol* 2012;72(4):536-49. **IF 11,193**

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Kraemer N, Morris-Rosendahl D, Kaindl AM (2012) Primäre autosomal-rezessive Mikrozephalie (MCPH). *Neuropaed in Klinik und Praxis.* Im Druck.

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Logan CV, Lucke B, Pottinger C, Abdelhamed ZA, Parry DA, Szymanska K, Diggle CP, van Riesen A, Morgan JE, Markham G, Ellis I, Manzur AY, Markham AF, Shires M, Helliwell T, Scoto M, Hübner C, Bonthron DT, Taylor GR, Sheridan E, Muntoni F, Carr IM, Schuelke M, Johnson CA. Mutations in *MEGF10*, a regulator of satellite cell myogenesis, cause early onset myopathy, areflexia, respiratory distress and dysphagia (EMARDD). *Nat Genet* 2011;43(12):1189-92. **IF 35,532**

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Dean JM, Wang X, Kaindl AM, Gressens P, Fleiss B, Hagberg H, Mallard C. Microglial MyD88 signaling regulates acute neuronal toxicity of LPS-stimulated microglia. *Brain Behav Immun* 2010;24(5):776-83. **IF 5,889**

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