Universitätsmedizin Göttingen
Publikationen und Hochschulschriften 2013
Neuropädiatrie

Journalbeiträge

1. Balint B, Haas J, Schwarz A, Jarius S, Fürwentsches A, Engelhardt K, Bussmann C, Ebinger F, Fritzsching B, Paul F, Seidel U, Vlaho S, Huppke P, Gärtner J, Wildemann B (2013) T-cell homeostasis in pediatric multiple sclerosis: old cells in young patients. NEUROLOGY, 81(9): 784-92.
2. Banne E, Atawneh O, Henneke M, Brockmann K, Gärtner J, Elpeleg O, Edvardson S (2013) West syndrome, microcephaly, grey matter heterotopia and hypoplasia of corpus callosum due to a novel ARFGEF2 mutation. J MED GENET, 50(11): 772-5.
3. Brendel C, Mielke B, Hillebrand M, Gärtner J, Huppke P (2013) Methotrexate treatment of FraX fibroblasts results in FMR1 transcription but not in detectable FMR1 protein levels. J NEURODEV DISORD, 5(1): 23.
4. Brockmann K (2013) Episodic movement disorders: from phenotype to genotype and back. CURR NEUROL NEUROSCI, 13(10): 379.
5. Buchert R, Uebe S, Radwan F, Tawamie H, Issa S, Shimazaki H, Henneke M, Ekici AB, Reis A, Abou Jamra R (2013) Mutations in the mitochondrial gene C12ORF65 lead to syndromic autosomal recessive intellectual disability and show genotype phenotype correlation. EUR J MED GENET, 56(11): 599-602.
6. Dreha-Kulaczewski S, Kalscheuer V, Tzschach A, Hu H, Helms G, Brockmann K, Weddige A, Dechent P, Schlüter G, Krätzner R, Ropers HH, Gärtner J, Zirn B (2013) A Novel SLC6A8 Mutation in a Large Family with X-Linked Intellectual Disability: Clinical and Proton Magnetic Resonance Spectroscopy Data of Both Hemizygous Males and Heterozygous Females. JIMD Rep, 11: -.
7. Grapp M, Wrede A, Schweizer M, Hüwel S, Galla HJ, Snaidero N, Simons M, Bückers J, Low PS, Urlaub H, Gärtner J, Steinfeld R (2013) Choroid plexus transcytosis and exosome shuttling deliver folate into brain parenchyma. NAT COMMUN, 4: 2123.
8. Grünert SC, Müllerleile S, De Silva L, Barth M, Walter M, Walter K, Meissner T, Lindner M, Ensenauer R, Santer R, Bodamer OA, Baumgartner MR, Brunner-Krainz M, Karall D, Haase C, Knerr I, Marquardt T, Hennermann JB, Steinfeld R, Beblo S, Koch HG, Konstantopoulou V, Scholl-Bürgi S, van Teeffelen-Heithoff A, Suormala T, Sperl W, Kraus JP, Superti-Furga A, Schwab KO, Sass JO (2013) Propionic acidemia: clinical course and outcome in 55 pediatric and adolescent patients. ORPHANET J RARE DIS, 8: 6.
9. Hermanns P, Grasberger H, Cohen R, Freiberg C, Dörr HG, Refetoff S, Pohlenz J (2013) Two cases of thyroid dysgenesis caused by different novel PAX8 mutations in the DNA-binding region: in vitro studies reveal different pathogenic mechanisms. THYROID, 23(7): 791-6.
10. Hülper P, Veszelka S, Walter FR, Wolburg H, Fallier-Becker P, Piontek J, Blasig IE, Lakomek M, Kugler W, Deli MA (2013) Acute effects of short-chain alkylglycerols on blood-brain barrier properties of cultured brain endothelial cells. BRIT J PHARMACOL, 169(7): 1561-73.
11. Hummel HM, Brück W, Dreha-Kulaczewski S, Gärtner J, Wuerfel J (2013) Pediatric onset multiple sclerosis: McDonald criteria 2010 and the contribution of spinal cord MRI. MULT SCLER J, 19(10): 1330-5.
12. Huppke P, Rostasy K, Karenfort M, Huppke B, Seidl R, Leiz S, Reindl M, Gärtner J (2013) Acute disseminated encephalomyelitis followed by recurrent or monophasic optic neuritis in pediatric patients. MULT SCLER J, 19(7): 941-6.
13. Koziolek M, Mühlhausen J, Friede T, Ellenberger D, Sigler M, Huppke B, Gärtner J, Müller GA, Huppke P (2013) Therapeutic apheresis in pediatric patients with acute CNS inflammatory demyelinating disease. BLOOD PURIFICAT, 36(2): 92-7.
14. Krause C, Rosewich H, Woehler A, Gärtner J (2013) Functional analysis of PEX13 mutation in a Zellweger syndrome spectrum patient reveals novel homooligomerization of PEX13 and its role in human peroxisome biogenesis. HUM MOL GENET, 22(19): 3844-57.
15. Krauth C, Liersch S, Sterdt E, Henze V, Röbl M, Walter U (2013) [Health economic evaluation of health promotion - the example "fit for pisa"]. GESUNDHEITSWESEN, 75(11): 742-6.
16. Neunhöffer H, Gold A, Hoerauf H, Herbort C, Heiligenhaus A, Zimmermann O, Feltgen N (2013) Isolated ocular Jarisch-Herxheimer reaction after initiating tuberculostatic therapy in a child. Int Ophthalmol, online: online.
17. Pfeifenbring S, von Baumgarten L, Schüller U, Rosewich H, Thal DR, Wirtz CR, Hecht M, Terpolilli NA, Brück W (2013) Biopsy findings of symptomatic cerebral X-linked adrenoleukodystrophy and histological differentiation from multiple sclerosis. NEUROPATH APPL NEURO, 1: 1-10.

18. Ravenscroft G, Miyatake S, Lehtokari VL, Todd EJ, Vornanen P, Yau KS, Hayashi YK, Miyake N, Tsurusaki Y, Doi H, Saitsu H, Osaka H, Yamashita S, Ohya T, Sakamoto Y, Koshimizu E, Imamura S, Yamashita M, Ogata K, Shiina M, Bryson-Richardson RJ, Vaz R, Ceyhan O, Brownstein CA, Swanson LC, Monnot S, Romero NB, Amthor H, Kresoje N, Sivadorai P, Kiraly-Borri C, Haliloglu G, Talim B, Orhan D, Kale G, Charles AK, Fabian VA, Davis MR, Lammens M, Sewry CA, Manzur A, Muntoni F, Clarke NF, North KN, Bertini E, Nevo Y, Willichowski E, Silberg IE, Topaloglu H, Beggs AH, Allcock RJN, Nishino I, Wallgren-Pettersson C, Matsumoto N, Laing NG (2013) Mutations in KLHL40 are a frequent cause of severe autosomal-recessive nemaline myopathy. AM J HUM GENET, 93(1): 6-18.
19. Rensing-Ehl A, Janda A, Lorenz MR, Gladstone BP, Fuchs I, Abinun M, Albert M, Butler K, Cant A, Cseh AM, Ebinger M, Goldacker S, Hambleton S, Hebart H, Houet L, Kentouche K, Kühnle I, Lehmberg K, Mejstrikova E, Niemeyer C, Minkov M, Neth O, Dückers G, Owens S, Rösler J, Schilling FH, Schuster V, Seidel MG, Smisek P, Sukova M, Svec P, Wiesel T, Gathmann B, Schwarz K, Vach W, Ehl S, Speckmann C (2013) Sequential decisions on FAS sequencing guided by biomarkers in patients with lymphoproliferation and autoimmune cytopenia. HAEMATOLOGICA, 98(12): 1948-55.
20. Röbl M, de Souza M, Schiel R, Gellhaus I, Zwiauer K, Holl RW, Wiegand S (2013) The key role of psychosocial risk on therapeutic outcome in obese children and adolescents. Results from a longitudinal multicenter study. OBESITY FACTS, 6(3): 297-305.
21. Rudnik-Schöneborn S, Senderek J, Jen JC, Houge G, Seeman P, Puchmajerová A, Graul-Neumann L, Seidel U, Korinthenberg R, Kirschner J, Seeger J, Ryan MM, Muntoni F, Steinlin M, Sztriha L, Colomer J, Hübner C, Brockmann K, Van Maldergem L, Schiff M, Holzinger A, Barth P, Reardon W, Yourshaw M, Nelson SF, Eggermann T, Zerres K (2013) Pontocerebellar hypoplasia type 1: clinical spectrum and relevance of EXOSC3 mutations. NEUROLOGY, 80(5): 438-46.
22. Schäfer A, Gratchev A, Seebode C, Hofmann L, Schubert S, Laspe P, Apel A, Ohlenbusch A, Tzvetkov M, Weishaupt C, Oji V, Schön MP, Emmert S (2013) Functional and molecular genetic analyses of nine newly identified XPD-deficient patients reveal a novel mutation resulting in TTD as well as in XP/CS complex phenotypes. EXP DERMATOL, 22(7): 486-9.
23. Schäfer A, Hofmann L, Gratchev A, Laspe P, Schubert S, Schürer A, Ohlenbusch A, Tzvetkov M, Hallermann C, Reichrath J, Schön MP, Emmert S (2013) Molecular genetic analysis of 16 XP-C patients from Germany: environmental factors predominately contribute to phenotype variations. EXP DERMATOL, 22(1): 24-9.
24. Schäfer A, Schubert S, Gratchev A, Seebode C, Apel A, Laspe P, Hofmann L, Ohlenbusch A, Mori T, Kobayashi N, Schürer A, Schön MP, Emmert S (2013) Characterization of three XPG-defective patients identifies three missense mutations that impair repair and transcription. J INVEST DERMATOL, 133(7): 1841-9.
25. Schlotawa L, Hotz A, Zeschnigk C, Hartmann B, Gärtner J, Morris-Rosendahl D (2013) Cerebellar ataxia, mental retardation and dysequilibrium syndrome 1 (CAMRQ1) caused by an unusual constellation of VLDLR mutation. J NEUROL, 260(6): 1678-80.
26. Schlotawa L, Radhakrishnan K, Baumgartner M, Schmid R, Schmidt B, Dierks T, Gärtner J (2013) Rapid degradation of an active formylglycine generating enzyme variant leads to a late infantile severe form of multiple sulfatase deficiency. EUR J HUM GENET, 21(9): 1020-3.
27. Schmidt H, Kretzschmar B, Lingor P, Pauli S, Schramm P, Otto M, Ohlenbusch A, Brockmann K (2013) Acute onset of adult Alexander disease. J NEUROL SCI, 331(1-2): 152-4.
28. Schwaibold EMC, Zoll B, Burfeind P, Hobbiebrunken E, Wilken B, Funke R, Shoukier M (2013) A 3p interstitial deletion in two monozygotic twin brothers and an 18-year-old man: further characterization and review. AM J MED GENET A, 161(10): 2634-40.
29. Shoukier M, Klein N, Auber B, Wickert J, Schröder J, Zoll B, Burfeind P, Bartels I, Alsat EA, Lingen M, Grzmil P, Schulze S, Keyser J, Weise D, Borchers M, Hobbiebrunken E, Röbl M, Gärtner J, Brockmann K, Zirn B (2013) Array CGH in patients with developmental delay or intellectual disability: are there phenotypic clues to pathogenic copy number variants? CLIN GENET, 83(1): 53-65.
30. Shoukier M, Fuchs S, Schwaibold E, Lingen M, Gärtner J, Brockmann K, Zirn B (2013) Microduplication of 3p26.3 in nonsyndromic intellectual disability indicates an important role of CHL1 for normal cognitive function. NEUROPEDIATRICS, 44(5): 268-71.

31. Zhao XW, Gazendam RP, Drewniak A, van Houdt M, Tool ATJ, van Hamme JL, Kustiawan I, Meijer AB, Janssen H, Russell DG, van de Corput L, Tesselaar K, Boelens JJ, Kuhnle I, Van Der Werff Ten Bosch J, Kuijpers TW, van den Berg TK (2013) Defects in neutrophil granule mobilization and bactericidal activity in familial hemophagocytic lymphohistiocytosis type 5 (FHL-5) syndrome caused by STXBP2/Munc18-2 mutations. BLOOD, 122(1): 109-11.
32. Zhukova N, Ramaswamy V, Remke M, Pfaff E, Shih DJH, Martin DC, Castelo-Branco P, Baskin B, Ray PN, Bouffet E, von Bueren AO, Jones DTW, Northcott PA, Kool M, Sturm D, Pugh TJ, Pomeroy SL, Cho YJ, Pietsch T, Gessi M, Rutkowski S, Bognar L, Klekner A, Cho BK, Kim SK, Wang KC, Eberhart CG, Fevre-Montange M, Fouladi M, French PJ, Kros M, Grajkowska WA, Gupta N, Weiss WA, Hauser P, Jabado N, Jouvet A, Jung S, Kumabe T, Lach B, Leonard JR, Rubin JB, Liau LM, Massimi L, Pollack IF, Shin Ra Y, Van Meir EG, Zitterbart K, Schüller U, Hill RM, Lindsey JC, Schwalbe EC, Bailey S, Ellison DW, Hawkins C, Malkin D, Clifford SC, Korshunov A, Pfister S, Taylor MD, Tabori U (2013) Subgroup-specific prognostic implications of TP53 mutation in medulloblastoma. J CLIN ONCOL, 31(23): 2927-35.

Habilitationen

1. Thoms S (2013) Peroxisomenbiogenese. Habilitation Universität Göttingen.

Medizinische Dissertationen

1. Just IA, Dr. med. (2013) Charakterisierung neuer Mutationen im FOLRI1-Gen. Dissertation Universität Göttingen.