

Journalbeiträge

1. Bechtold S, Blaschek A, Raile K, Dost A, Freiberg C, Askenas M, Fröhlich-Reiterer E, Molz E, Holl RW (2014) Higher relative risk for multiple sclerosis in a pediatric and adolescent diabetic population: analysis from DPV database. DIABETES CARE 37(1): 96-101, doi: 10.2337/dc13-1414
2. Cohen Y, Klug YA, Dimitrov L, Erez Z, Chuartzman SG, Elninger D, Yofe I, Soliman K, Gärtner J, Thoms S, Schekman R, Elbaz-Alon Y, Zalckvar E, Schuldiner M (2014) Peroxisomes are juxtaposed to strategic sites on mitochondria. Mol Biosyst 10(7): 1742-8, doi: 10.1039/c4mb00001c
3. Dieks JK, Baumer A, Wilichowski E, Rauch A, Sigler M (2014) Microcephalic osteodysplastic primordial dwarfism type II (MOPD II) with multiple vascular complications misdiagnosed as Dubowitz syndrome. EUR J PEDIATR 173(9): 1253-6, doi: 10.1007/s00431-014-2368-5
4. Distelmaier F, Huppke P, Pieperhoff P, Amunts K, Schaper J, Morava E, Mayatepek E, Kohlhase J, Karenfort M (2014) Biotin-responsive Basal Ganglia disease: a treatable differential diagnosis of leigh syndrome. JIMD Rep 13: 53-7, doi: 10.1007/8904_2013_271
5. Haack TB, Gorza M, Danhauser K, Mayr JA, Haberberger B, Wieland T, Kremer L, Strecker V, Graf E, Memari Y, Ahting U, Kopajtich R, Wortmann SB, Rodenburg RJ, Kotzaeridou U, Hoffmann GF, Sperl W, Wittig I, Wilichowski E, Schottmann G, Schuelke M, Plecko B, Stephani U, Strom TM, Meitinger T, Prokisch H, Freisinger P (2014) Phenotypic spectrum of eleven patients and five novel MTFMT mutations identified by exome sequencing and candidate gene screening. MOL GENET METAB 111(3): 342-52, doi: 10.1016/j.ymgme.2013.12.010
6. Huppke B, Ellenberger D, Rosewich H, Friede T, Gärtner J, Huppke P (2014) Clinical presentation of pediatric multiple sclerosis before puberty. EUR J NEUROL 21(3): 441-6, doi: 10.1111/ene.12327
7. Huppke P, Hummel H, Ellenberger D, Pfeifenbring S, Stark W, Huppke B, Brück W, Gärtner J (2014) JC virus antibody status in a pediatric multiple sclerosis cohort: Prevalence, conversion rate and influence on disease severity. MULT SCLER J 21(4): 382-7, doi: 10.1177/1352458514543340
8. Huppke P, Wegener E, Böhrer-Rabel H, Bolz HJ, Zoll B, Gärtner J, Bergmann C (2014) Tectonic gene mutations in patients with Joubert syndrome. EUR J HUM GENET 9(12): 1-14, doi: 10.1038/ejhg.2014.160
9. Kehrer C, Groeschel S, Kustermann-Kuhn B, Bürger F, Köhler W, Kohlschütter A, Bley A, Steinfeld R, Gieselmann V, Krägeloh-Mann I, German LEUKONET (2014) Language and cognition in children with metachromatic leukodystrophy: onset and natural course in a nationwide cohort. ORPHANET J RARE DIS 9: 18, doi: 10.1186/1750-1172-9-18
10. Kirschner J, Schorling D, Hauschke D, Rensing-Zimmermann C, Wein U, Grieben U, Schottmann G, Schara U, Konrad K, Müller-Felber W, Thiele S, Wilichowski E, Hobbiebrunken E, Stettner GM, Korinthenberg R (2014) Somatotropin treatment of spinal muscular atrophy: a placebo-controlled, double-blind crossover pilot study. NEUROMUSCULAR DISORD 24(2): 134-42, doi: 10.1016/j.nmd.2013.10.011
11. Lehmann J, Schubert S, Schäfer A, Apel A, Laspe P, Schiller S, Ohlenbusch A, Gratchev A, Emmert S (2014) An unusual mutation in the XPG gene leads to an internal in-frame deletion and a XP/CS complex phenotype. BRIT J DERMATOL 171(4): 903-5, doi: 10.1111/bjd.13035
12. Marttila M, Lehtokari VL, Marston S, Nyman TA, Barnerias C, Beggs AH, Bertini E, Ceyhan-Birsoy O, Cintas P, Gerard M, Gilbert-Dussardier B, Hogue JS, Longman C, Eymard B, Frydman M, Kang PB, Klinge L, Kolski H, Lochmüller H, Magy L, Manel V, Mayer M, Mercuri E, North KN, Peudenier-Robert S, Pihko H, Probst FJ, Reisin R, Stewart W, Taratuto AL, de Visser M, Wilichowski E, Winer J, Nowak K, Laing NG, Winder TL, Monnier N, Clarke NF, Pelin K, Grönholm M, Wallgren-Pettersson C (2014) Mutation update and genotype-phenotype correlations of novel and previously described mutations in TPM2 and TPM3 causing congenital myopathies. HUM MUTAT 35(7): 779-90, doi: 10.1002/humu.22554
13. Menke J, Pauli S, Sigler M, Kühnle I, Shoukier M, Zoll B, Ganster C, Salinas-Riester G, Schaefer IM (2014) Uniparental Trisomy of a Mutated HRAS Proto-Oncogene in Embryonal Rhabdomyosarcoma of a Patient With Costello Syndrome. J CLIN ONCOL 33(13): e62-e65, doi: 10.1200/JCO.2013.49.6539
14. Na U, Yu W, Cox J, Bricker DK, Brockmann K, Rutter J, Thummel CS, Winge DR (2014) The LYR factors SDHAF1 and SDHAF3 mediate maturation of the iron-sulfur subunit of succinate dehydrogenase. CELL METAB 20(2): 253-66, doi: 10.1016/j.cmet.2014.05.014
15. Nishri D, Edvardson S, Lev D, Leshinsky-Silver E, Ben-Sira L, Henneke M, Lerman-Sagie T, Blumkin L (2014) Diagnosis by whole exome sequencing of atypical infantile onset Alexander disease masquerading as a mitochondrial disorder. EUR J PAEDIATR NEURO 18(4): 495-501, doi: 10.1016/j.ejpn.2014.03.009
16. Reinhardt K, Weiss S, Rosenbauer J, Gärtner J, von Kries R (2014) Multiple sclerosis in children and adolescents: incidence and clinical picture - new insights from the nationwide German surveillance (2009-2011). EUR J NEUROL 21(4): 654-9, doi: 10.1111/ene.12371
17. Rosewich H, Baethmann M, Ohlenbusch A, Gärtner J, Brockmann K (2014) A novel ATP1A3 mutation with unique clinical presentation. J NEUROL SCI 341(1-2): 133-5, doi: 10.1016/j.jns.2014.03.034

18. Rosewich H, Ohlenbusch A, Huppke P, Schlotawa L, Baethmann M, Carrilho I, Fiori S, Lourenço CM, Sawyer S, Steinfeld R, Gärtner J, Brockmann K (2014) The expanding clinical and genetic spectrum of ATP1A3-related disorders. *NEUROLOGY* 82(11): 945-55, doi: 10.1212/WNL.0000000000000212
19. Rosewich H, Waterham H, Poll-The BT, Ohlenbusch A, Gärtner J (2014) Clinical utility gene card for: Zellweger syndrome spectrum. *EUR J HUM GENET* e1-e4: 1-4, doi: 10.1038/ejhg.2014.250
20. Rosewich H, Weise D, Ohlenbusch A, Gärtner J, Brockmann K (2014) Phenotypic overlap of alternating hemiplegia of childhood and CAPOS syndrome. *NEUROLOGY* 83(9): 861-3, doi: 10.1212/WNL.0000000000000735
21. Schueren F, Lingner T, George R, Hofhuis J, Dickel C, Gärtner J, Thoms S (2014) Peroxisomal lactate dehydrogenase is generated by translational readthrough in mammals. *ELIFE* 3: e03640, doi: 10.7554/elife.03640
22. Schulz Y, Freese L, Mänz J, Zoll B, Völter C, Brockmann K, Bögershausen N, Becker J, Wollnik B, Pauli S (2014) CHARGE and Kabuki syndromes: a phenotypic and molecular link. *HUM MOL GENET* 23(16): 4396-405, doi: 10.1093/hmg/ddu156
23. Schwaibold EMC, Smogavec M, Hobbiebrunken E, Winter L, Zoll B, Burfeind P, Brockmann K, Pauli S (2014) Intragenic duplication of EHMT1 gene results in Kleefstra syndrome. *MOL CYTOGENET* 7(1): 74, doi: 10.1186/s13039-014-0074-7
24. Schweingruber N, Fischer HJ, Fischer L, van den Brandt J, Karabinskaya A, Labi V, Villunger A, Kretzschmar B, Huppke P, Simons M, Tuckermann JP, Flügel A, Lühder F, Reichardt HM (2014) Chemokine-mediated redirection of T cells constitutes a critical mechanism of glucocorticoid therapy in autoimmune CNS responses. *ACTA NEUROPATHOL* 127(5): 713-29, doi: 10.1007/s00401-014-1248-4
25. Sidhu NS, Schreiber K, Pröpper K, Becker S, Usón I, Sheldrick GM, Gärtner J, Krätzner R, Steinfeld R (2014) Structure of sulfamidase provides insight into the molecular pathology of mucopolysaccharidosis IIIA. *ACTA CRYSTALLOGR D* 70(Pt 5): 1321-35, doi: 10.1107/S1399004714002739
26. Spiegler S, Najm J, Liu J, Gkalympoudis S, Schröder W, Borck G, Brockmann K, Elbracht M, Fauth C, Ferbert A, Freudenberg L, Grasshoff U, Hellenbroich Y, Henn W, Hoffjan S, Hüning I, Korenke GC, Kroisel PM, Kunstmänn E, Mair M, Munk-Schulenburg S, Nikouashman O, Pauli S, Rudnik-Schöneborn S, Sudholt I, Sure U, Tinschert S, Wiednig M, Zoll B, Ginsberg MH, Felbor U (2014) High mutation detection rates in cerebral cavernous malformation upon stringent inclusion criteria: one-third of probands are minors. *Mol Genet Genomic Med* 2(2): 176-85, doi: 10.1002/mgg3.60
27. Stockler S, Corvera S, Lambright D, Fogarty K, Nosova E, Leonard D, Steinfeld R, Ackerley C, Shyr C, Au N, Selby K, van Allen M, Vallance H, Wevers R, Watkins D, Rosenblatt D, Ross CJ, Conibear E, Wasserman W, van Karnebeek C (2014) Single point mutation in Rabenosyn-5 in a female with intractable seizures and evidence of defective endocytotic trafficking. *ORPHANET J RARE DIS* 9(1): 141, doi: 10.1186/s13023-014-0141-5
28. Synofzik M, Haack TB, Kopajtich R, Gorza M, Rapaport D, Greiner M, Schönfeld C, Freiberg C, Schorr S, Holl RW, Gonzalez MA, Fritzsche A, Fallier-Becker P, Zimmermann R, Strom TM, Meitinger T, Züchner S, Schüle R, Schöls L, Prokisch H (2014) Absence of BiP co-chaperone DNAJC3 causes diabetes mellitus and multisystemic neurodegeneration. *AM J HUM GENET* 95(6): 689-97, doi: 10.1016/j.ajhg.2014.10.013
29. Thorwarth A, Schnittert-Hübener S, Schrumpf P, Müller I, Jyrch S, Dame C, Biebermann H, Kleinau G, Katchanov J, Schuelke M, Ebert G, Steininger A, Bönnemann C, Brockmann K, Christen HJ, Crock P, deZegher F, Griese M, Hewitt J, Ivarsson S, Hübner C, Kapelari K, Plecko B, Rating D, Stoeva I, Ropers HH, Grüters A, Ullmann R, Krude H (2014) Comprehensive genotyping and clinical characterisation reveal 27 novel NKX2-1 mutations and expand the phenotypic spectrum. *J MED GENET* 51(6): 375-87, doi: 10.1136/jmedgenet-2013-102248
30. Volgin DV, Lu JW, Stettner GM, Mann GL, Ross RJ, Morrison AR, Kubin L (2014) Time- and behavioral state-dependent changes in posterior hypothalamic GABA_A receptors contribute to the regulation of sleep. *PLOS ONE* 9(1): e86545, doi: 10.1371/journal.pone.0086545
31. Wegener E, Brendel C, Fischer A, Hülsmann S, Gärtner J, Huppke P (2014) Characterization of the MeCP2R168X knockin mouse model for Rett syndrome. *PLOS ONE* 9(12): e115444, doi: 10.1371/journal.pone.0115444
32. Wiegand S, Keller KM, Lob-Corzilius T, Pott W, Reinehr T, Röbl M, Stachow R, Tuschy S, Weidanz I, Widhalm K, de Zwaan M, Holl RW (2014) Predicting weight loss and maintenance in overweight/obese pediatric patients. *Horm Res Paediatr* 82(6): 380-7, doi: 10.1159/000368963
33. Wuerfel E, Bien CG, Vincent A, Woodhall M, Brockmann K (2014) Glycine receptor antibodies in a boy with focal epilepsy and episodic behavioral disorder. *J NEUROL SCI* 343(1-2): 180-2, doi: 10.1016/j.jns.2014.05.014
34. Zou Y, Zwolanek D, Izu Y, Gandhy S, Schreiber G, Brockmann K, Devoto M, Tian Z, Hu Y, Veit G, Meier M, Stetefeld J, Hicks D, Straub V, Voermans NC, Birk DE, Barton ER, Koch M, Bönnemann CG (2014) Recessive and dominant mutations in COL12A1 cause a novel EDS/myopathy overlap syndrome in humans and mice. *HUM MOL GENET* 23(9): 2339-52, doi: 10.1093/hmg/ddt627

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Buchbeiträge

1. Gärtner J, Rosewich H (2014) Peroxisomale Krankheiten. In: Hoffmann GF, Lenze MJ, Spranger J, Zepp F (Hrsg.) Pädiatrie - Grundlagen und Praxis. Springer, 563-569
2. Gärtner J (2014) Neurometabolische Erkrankungen. In: Reinhardt D, Nicolai T, Zimmer, KP (Hrsg.) Therapie der Krankheiten im Kindes- und Jugendalter. Springer, 1185-1190
3. Hollak C, Kettwig M, Schlotawa L, Steinfeld R (2014) Lysosomal Storage Disorders Including Neuronal Ceroid Lipofuscinoses. In: Blau, N., Duran, M., Gibson, K.M., Dionisi-Vici, C (Hrsg.) Physician's Guide to the Diagnosis, Treatment, and Follow-Up of Inherited Metabolic Diseases. Springer, 399-437
4. Rosewich H, Gärtner J (2014) Peroxisomale Störungen. In: Dahl S, Lammert F, Ullrich K, Wendel U (Hrsg.) Angeborene Stoffwechselkrankheiten bei Erwachsenen. Springer, 449-460
5. 3. Gärtner J, Huppke P (2014) Multiple Sklerose (MS) und MS ähnliche Erkrankungen. In: Hoffmann GF, Lenze MJ, Spranger J, Zepp F (Hrsg.) Pädiatrie - Grundlagen und Praxis. Springer, 1747-1751

Habilitationen

1. Rosewich H (2014) Klinische und genetische Charakterisierung ATP1A3-assozierter Erkrankungen. Habilitation Universität Göttingen.
2. Stettner GM (2014) Die zentrale Kontrolle der oberen Atemwege und deren neuroplastische Adaptation nach chronisch intermittierender Hypoxie. Habilitation Universität Göttingen.