

Abteilung "Pädiatrie II mit Schwerpunkt Neuropädiatrie"

Journalbeiträge

1. Archer HL, Whatley SD, Evans JC, Ravine D, Huppke P, Bunyan D, Kerr AM, Kerr B, Sweeney E, Davies SJ, Reardon W, Horn J, Macdermot KD, Smith RA, Magee A, Donaldson A, Crow Y, Hermon G, Miedzybrodzka Z, Cooper DN, Lazarou L, Butler R, Sampson JR, Pilz DT, Laccone F, Clarke AJ (2005) Gross rearrangements of the MECP2 gene are found in both classical and atypical Rett Syndrome. *J MED GENET*, 43(5): 451-6.
2. Blaes F, Fühlhuber V, Korfei M, Tschernatsch M, Behnisch W, Rostasy K, Hero B, Kaps M, Preissner KT (2005) Surface-binding autoantibodies to cerebellar neurons in opsoclonus syndrome. *ANN NEUROL*, 58(2): 313-7.
3. Brockmann K (2005) Tarsaltunnel-Syndrom bei einem 7jährigen Jungen. *Kinderärztl Prax*, 1: 32-35.
4. Brockmann K (2005) Paroxysmale Dyskinesien und globale Retardierung bei Knaben mit Defizienz des Schilddrüsenhormon-Transporters MCT8. *Kinderärztl Prax*, 1: 36-41.
5. Brockmann K, Dumitrescu AM, Best TT, Hanefeld F, Refetoff S (2005) X-linked paroxysmal dyskinesia and severe global retardation caused by defective MCT8 gene. *J NEUROL*, 252(6): 663-6.
6. Brockmann K, Huppke P, Karenfort M, Gärtner J, Höger C (2005) Visually self-induced seizures sensitive to round objects. *EPILEPSIA*, 46(5): 786-9.
7. Brockmann K, Simpson MA, Faber A, Bönnemann C, Crosby AH, Gärtner J (2005) Complicated hereditary spastic paraplegia with thin corpus callosum (HSP-TCC) and childhood onset. *NEUROPEDIATRICS*, 36(4): 274-8.
8. Brockmann K, Stolpe S, Fels C, Khan N, Kulozik AE, Pekrun A (2005) Moyamoya syndrome associated with hemolytic anemia due to Hb Alesha. *J PEDIAT HEMATOL ONC*, 27(8): 436-40.
9. Diepold K, Schütz B, Rostasy K, Wilken B, Hougaard P, Güttler F, Romstad A, Birk Møller L (2005) Levodopa-responsive infantile parkinsonism due to a novel mutation in the tyrosine hydroxylase gene and exacerbation by viral infections. *MOVEMENT DISORD*, 20(6): 764-7.
10. Eiffert H, Karsten A, Ritter K, Ohlenbusch A, Schlott T, Laskawi R, Christen HJ (2005) Autoantibodies to human manganese superoxide dismutase (MnSOD) in children with facial palsy due to neuroborreliosis. *NEUROPEDIATRICS*, 36(6): 386-8.
11. Elleder M, Jerábková M, Befekadu A, Hřebíček M, Berná L, Ledvinová J, H Iková H, Rosewich H, Schymik N, Paton BC, Harzer K (2005) Prosaposin deficiency -- a rarely diagnosed, rapidly progressing, neonatal neurovisceral lipid storage disease. Report of a further patient. *NEUROPEDIATRICS*, 36(3): 171-80.
12. Hanefeld FA, Brockmann K, Pouwels PJ, Wilken B, Frahm J, Dechent P (2005) Quantitative proton MRS of Pelizaeus-Merzbacher disease: evidence of dys- and hypomyelination. *NEUROLOGY*, 65(5): 701-6.
13. Henneke M, Preuss N, Engelbrecht V, Aksu F, Bertini E, Bibat G, Brockmann K, Hübner C, Mayer M, Mejaski-Bosnjak V, Naidu S, Neumaier-Probst E, Rodriguez D, Weisz W, Kohlschütter A, Gärtner J (2005) Cystic leukoencephalopathy without megalencephaly: a distinct disease entity in 15 children. *NEUROLOGY*, 64(8): 1411-6.
14. Henneke M, Abicht A, Müller JS, Schara U, Gärtner J, Brockmann K (2005) Der interessante Fall: ein 20 monatiger Junge mit motorischer Retardierung und fluktuierender Ptose. *Kinderärztl Prax*, 5: 314-315.
15. Huppke B, Huppke P (2005) Der diagnostische Blick. *Kinderärztl Prax*, 6: 396.
16. Huppke P, Gärtner J (2005) Molecular diagnosis of Rett syndrome. *J CHILD NEUROL*, 20(9): 732-6.
17. Huppke P, Kallenberg K, Gärtner J (2005) Perisylvian polymicrogyria in Landau-Kleffner syndrome. *NEUROLOGY*, 64(9): 1660.
18. Huppke P, Ohlenbusch A, Brendel C, Laccone F, Gärtner J (2005) Mutation analysis of the HDAC 1, 2, 8 and CDKL5 genes in Rett syndrome patients without mutations in MECP2. *AM J MED GENET A*, 137(2): 136-8.
19. Jerkic S, Rosewich H, Scharf JG, Perske C, Füzesi L, Wilichowski E, Gärtner J (2005) Colorectal cancer in two pre-teenage siblings with familial adenomatous polyposis. *EUR J PEDIATR*, 164(5): 306-10.
20. Kasper D, Planells-Cases R, Fuhrmann JC, Scheel O, Zeitz O, Ruether K, Schmitt A, Poët M, Steinfeld R, Schweizer M, Kornak U, Jentsch TJ (2005) Loss of the chloride channel CIC-7 leads to lysosomal storage disease and neurodegeneration. *EMBO J*, 24(5): 1079-91.
21. Klusmann A, Heinrich B, Stöpler H, Gärtner J, Mayatepek E, Von Kries R (2005) A decreasing rate of neural tube defects following the recommendations for periconceptional folic acid supplementation. *ACTA PAEDIATR*, 94(11): 1538-42.
22. Lüthje S, Zappel H, Schweigerer L, Wilichowski E (2005) Chronische Obstipation als Leitsymptom bei einem 6jährigen Patienten mit myotoner Dystrophie. *Kinderärztl Prax*, 4: 232-234.
23. Navia BA, Rostasy K (2005) The AIDS dementia complex: clinical and basic neuroscience with implications for novel molecular therapies. *NEUROTOX RES*, 8(1-2): 3-24.

Abteilung "Pädiatrie II mit Schwerpunkt Neuropädiatrie"

24. Ohlenbusch A, Henneke M, Brockmann K, Goerg M, Hanefeld F, Kohlschütter A, Gärtner J (2005) Identification of ten novel mutations in patients with eIF2B-related disorders. *Hum Mutat* (Internet-Ausgabe), 25(4): 411.
25. Pohl D, Rostasy K, Gärtner J, Hanefeld F (2005) Treatment of early onset multiple sclerosis with subcutaneous interferon beta-1a. *NEUROLOGY*, 64(5): 888-90.
26. Pohl D, Rostasy K, Krone B, Hanefeld F (2005) Baló's concentric sclerosis associated with primary human herpesvirus 6 infection. *J NEUROL NEUROSUR PS*, 76(12): 1723-5.
27. Rosewich H, Ohlenbusch A, Gärtner J (2005) Genetic and clinical aspects of Zellweger spectrum patients with PEX1 mutations. *J MED GENET*, 42(9): e58.
28. Rostasy K (2005) Kinderzeichnungen bei Migräne. *Kinderärztl Prax*, 2: 88-91.
29. Rostasy K, Gorgun G, Kleyner Y, Garcia A, Kramer M, Melanson SM, Mathys JM, Yiannoutsos C, Skolnik PR, Navia BA (2005) Tumor necrosis factor alpha leads to increased cell surface expression of CXCR4 in SK-N-MC cells. *J NEUROVIROL*, 11(3): 247-55.
30. Rostasy K, Monti L, Lipton SA, Hedreen JC, Gonzalez RG, Navia BA (2005) HIV leucoencephalopathy and TNFalpha expression in neurones. *J NEUROL NEUROSUR PS*, 76(7): 960-4.
31. Rostasy K, Withut E, Pohl D, Lange P, Ciesielcyk B, Diem R, Gärtner J, Otto M (2005) Tau, phospho-tau, and S-100B in the cerebrospinal fluid of children with multiple sclerosis. *J CHILD NEUROL*, 20(10): 822-5.
32. Rostásy KM (2005) Inflammation and neuroaxonal injury in multiple sclerosis and AIDS dementia complex: implications for neuroprotective treatment. *NEUROPEDIATRICS*, 36(4): 230-9.
33. Rostásy KM, Huppke P, Beckers B, Brockmann K, Degenhardt V, Wesche B, König F, Gärtner J (2005) Acute motor and sensory axonal neuropathy (AMSAN) in a 15-year-old boy presenting with severe pain and distal muscle weakness. *NEUROPEDIATRICS*, 36(4): 260-4.
34. Siegert S, Bahn E, Kramer ML, Schulz-Schaeffer WJ, Hewett JW, Breakefield XO, Hedreen JC, Rostasy KM (2005) TorsinA expression is detectable in human infants as young as 4 weeks old. *DEV BRAIN RES*, 157(1): 19-26.
35. Walter U, Kramer S, Röbl M (2005) [Physical (in)activity in childhood and adolescence]. *DEUT MED WOCHENSCHR*, 130(50): 2876-8.
36. Weiss E, Albrecht CF, Herms J, Behnke-Mursch J, Pekrun A, Brockmann K, Hess CF (2005) Malignant ectomesenchymoma of the cerebrum. Case report and discussion of therapeutic options. *EUR J PEDIATR*, 164(6): 345-9.
37. Weller S, Cajigas I, Morrell J, Obie C, Steel G, Gould SJ, Valle D (2005) Alternative splicing suggests extended function of PEX26 in peroxisome biogenesis. *AM J HUM GENET*, 76(6): 987-1007.
38. Zavadáková P, Fowler B, Suormala T, Novotna Z, Mueller P, Hennermann JB, Zeman J, Vilaseca MA, Vilarinho L, Gutsche S, Wilichowski E, Horneff G, Kozich V (2005) cbIE type of homocystinuria due to methionine synthase reductase deficiency: functional correction by minigene expression. *HUM MUTAT*, 25(3): 239-47.

Buchbeiträge

1. Gärtner J (2005) Genetisch bedingte Stoffwechselerkrankungen. In: Wallesch CW (Hg.) *Neurologie. Diagnostik und Therapie in Klinik und Praxis*. Urban & Fischer Verlag, Jena, 861-869.
2. Hanefeld F, Brockmann K, Dechent P (2005) MR spectroscopy in pediatric white matter disease. In: Gillard J, Waldman A, Barker P (Hg.) *Clinical MR Imaging*. Cambridge University Press, Oxford, 755-777.
3. Steinfeld R, Baethmann M, Gärtner J (2005) New aspects of childhood neurometabolic disorders. In: Panteliadis CP, Korinthenberg R (Hg.) *Paediatric Neurology*. Thieme Verlag, Stuttgart, 399-417.

Medizinische Dissertationen

1. Ruppert S, Dr. med., Beeinträchtigung der Farbdiskrimination bei Kindern mit Aufmerksamkeitsdefizit-/Hyperaktivitätsstörungen (ADHS). Dissertation Universität Göttingen 2005.
2. Schlotawa L, Dr. med., Stimulierung des Aktivierenden Proteins- 1 und der Jun- NH2- terminalen Kinase in Endometriumkarzinomzellen durch das Analogon des Luteinisierenden Hormon-Releasing-Hormons Triptorelin. Dissertation Universität Göttingen 2005.