Vogelflug 2016 Literaturliste

GENETIK

DeLuca AP, Weed MC, Haas CM, Halder JA, Stone EM. Apparent Usher Syndrome Caused by the Combination of BBS1-Associated Retinitis Pigmentosa and *SLC26A4*-Associated Deafness. JAMA Ophthalmol 2015 Aug;133(8):967-8

Graf J, Hellenbroich Y, Veelken N, Figueroa KP, Wolff S, Pulst S, Brüggemann N. Two different genetic diseases in the same patient: Coincident, concomitant, or causally related? Mov Disord 2016 Feb 22. doi: 10.1002/mds.26539.

Komara M, John A, Suleiman J, Ali BR, Al-Gazali L. Clinical and molecular delineation of dysequilibrium syndrome type 2 and profound sensorineural hearing loss in an inbred Arab family. Am J Med Genet A 2016 Feb;170(2):540-3

Martin F, Kana V, Mori AC, Fischer D, Parkin N, Boltshauser E, Rushing EJ, Klein A. Neurofibromatosis type 1 (NF1) with an unusually severe phenotype due to digeny for NF1 and ryanodine receptor 1 associated myopathy. Eur J Pediatr 2014 Dec;173(12):1691-4

Pasquier L, Fradin M, Chérot E, Martin-Coignard D, Colin E, Journel H, Demurger F, Akloul L, Quélin C, Jauffret V, Lucas J, Belaud-Rotureau MA, Odent S, Jaillard S. Karyotype is not dead (yet)! Eur J Med Genet 2016 Jan;59(1):11-5

Sun K, Jiang P, Chan KC, Wong J, Cheng YK, Liang RH, Chan WK, Ma ES, Chan SL, Cheng SH, Chan RW, Tong YK, Ng SS, Wong RS, Hui DS, Leung TN, Leung TY, Lai PB, Chiu RW, Lo YM. Plasma DNA tissue mapping by genome-wide methylation sequencing for noninvasive prenatal, cancer, and transplantation assessments. Proc Natl Acad Sci U S A 2015 Oct 6;112(40):E5503-12

Vu PQ, Tian JJ, Sadun AA. Genetic Incidentaloma in Ophthalmology. JAMA Ophthalmol 2016 Feb 1;134(2):123-4

Wong FC, Lo YM. Prenatal Diagnosis Innovation: Genome Sequencing of Maternal Plasma. Annu Rev Med 2016 Jan 14;67:419-32

NEUROFIBROMATOSE TYP 1

Afridi SK, Leschziner GD, Ferner RE. Prevalence and clinical presentation of headache in a National Neurofibromatosis 1 Service and impact on quality of life. Am J Med Genet A 2015 Oct;167A(10):2282-5

Blanchard G, Lafforgue MP, Lion-François L, Kemlin I, Rodriguez D, Castelnau P, Carneiro M, Meyer P, Rivier F, Barbarot S, Chaix Y; NF France network. Systematic MRI in NF1 children under six years of age for the diagnosis of optic pathway gliomas. Study and outcome of a French cohort. Eur J Paediatr Neurol 2016 Mar;20(2):275-81

Caen S, Cassiman C, Legius E, Casteels I. Comparative study of the ophthalmological examinations in neurofibromatosis type 1. Proposal for a new screening algorithm. Eur J Paediatr Neurol 2015 Jul;19(4):415-22

Cornett KM, North KN, Rose KJ, Burns J. Muscle weakness in children with neurofibromatosis type 1. Dev Med Child Neurol 2015 Aug;57(8):733-6

Ejerskov C, Lasgaard M, Østergaard JR. Teenagers and young adults with neurofibromatosis type 1 are more likely to experience loneliness than siblings without the illness. Acta Paediatr 2015 Jun;104(6):604-9

Marque M, Roubertie A, Jaussent A, Carneiro M, Meunier L, Guillot B, Pinson L, Pinson S, Bessis D. Nevus anemicus in neurofibromatosis type 1: a potential new diagnostic criterion. J Am Acad Dermatol 2013 Nov;69(5):768-75

Vassen P, Rosenbaum T. Nevus anemicus as an additional diagnostic marker of neurofobromatosis type 1 in childhood. Neuropediatrics 2016, epub

NEUROIMAGING

Al-Hashim AH, Blaser S, Raybaud C, MacGregor D. Corpus callosum abnormalities: neuroradiological and clinical correlations. Dev Med Child Neurol 2015 Dec 9.

Bates JE, Almast J, Augustine EF. Neonatal dentate nucleus T2 hyperintensity after in utero metronidazole exposure. Neurology. 2015 Sep 15;85(11):1006

Booth TN, Rollins NK. Spectrum of Clinical and Associated MR Imaging Findings in Children with Olfactory Anomalies. Am J Neuroradiol 2016 Mar 17.

De Keersmaecker B, Ramaekers P, Claus F, Witters I, Ortibus E, Naulaers G, Van Calenbergh F, De Catte L. Outcome of 12 antenatally diagnosed fetal arachnoid cysts: case series and review of the literature. Eur J Paediatr Neurol 2015 Mar;19(2):114-21

McErlean A, Abdalla K, Donoghue V, Ryan S. The dentate nucleus in children: normal development and patterns of disease. Pediatr Radiol 2010 Mar;40(3):326-39

Mutch CA, Poduri A, Sahin M, Barry B, Walsh CA, Barkovich AJ. Disorders of Microtubule Function in Neurons: Imaging Correlates. Am J Neuroradiol 2016 Mar;37(3):528-35

Sundarakumar DK, Farley SA, Smith CM, Maravilla KR, Dighe MK, Nixon JN. Absent cavum septum pellucidum: a review with emphasis on associated commissural abnormalities. Pediatr Radiol 2015 Jul;45(7):950-64

Yoganathan S, Arunachal G, Sudhakar SV, Rajaraman V, Thomas M, Danda S. Beta Propellar Protein-Associated Neurodegeneration: A Rare Cause of Infantile Autistic Regression and Intracranial Calcification. Neuropediatrics 2016 Apr;47(2):123-7

Zuccoli G, Yannes MP, Nardone R, Bailey A, Goldstein A. Bilateral symmetrical basal ganglia and thalamic lesions in children: an update (2015). Neuroradiology 2015 Oct;57(10):973-8.

VARIA

Armangue T, Moris G, Cantarín-Extremera V, *et al.* Autoimmune post–herpes simplex encephalitis of adults and teenagers. *Neurology* 2015; **85**: 1736–43.

Giese AK, Mascher H, Grittner U, Eichler S, Kramp G, Lukas J, te Vruchte D, Al Eisa N, Cortina-Borja M, Porter FD, Platt FM, Rolfs A. A novel, highly sensitive and specific biomarker for Niemann-Pick type C1 disease. Orphanet J Rare Dis 2015 Jun 17;10:78

Graus F, Titulaer M, Balu R, *et al.* A clinical approach to diagnosis of autoimmune encephalitis. *The Lancet Neurology* 2016. doi:10.1016/S1474-4422(15)00401-9.

Gupta S, Mak S, Detsky AS. A piece of my mind. The other generation. JAMA. 2015 Apr 7;313(13):1319-20.

Hougaard A, Lindberg U, Arngrim N, *et al.* Evidence of a Christmas spirit network in the brain: functional MRI study. *BMJ* 2015; h6266.

Klinke G, Rohrbach M, Giugliani R, Burda P, Baumgartner MR, Tran C, Gautschi M, Mathis D, Hersberger M. LC-MS/MS based assay and reference intervals in children and adolescents for oxysterols elevated in Niemann-Pick diseases. Clin Biochem 2015 Jun;48(9):596-602

Ropper A. How to determine if you have succeeded at neurology residency. *Ann Neurol* 2016; **79**: 339–41.

Rutkove SB, Shefner JM, Bowser R, Benatar M. To travel or not to travel: The modern day struggle of the academic researcher. Ann Neurol 2015 Nov;78(5):667-9

MOVEMENT DISORDERS

Chen D-H, Méneret A, Friedman J, *et al.* ADCY5-related dyskinesia: Broader spectrum and genotype-phenotype correlations. *Neurology* 2015; **85**: 2026–35.

Schüle R, Wiethoff S, Martus P, *et al.* Hereditary spastic paraplegia: Clinicogenetic lessons from 608 patients. *Ann Neurol* 2016; **79**: 646–58.

Zutt R, Egmond M, Elting J, *et al.* A novel diagnostic approach to patients with myoclonus.*Nat Rev Neurology* 2015; **11**: 687–97. Tesson C, Koht J, Stevanin G. Delving into the complexity of hereditary spastic paraplegias: how unexpected phenotypes and inheritance modes are revolutionizing their nosology. *Human Genetics*2015; **134**: 511–38.

NEUROMETABOLISCHE ERKRANKUNGEN

Boczonadi V, Bansagi B, Horvath R. Reversible infantile mitochondrial diseases. *J Inherit Metab Dis*2015; **38**: 427–35.

Jaeger B, Bosch A. Clinical presentation and outcome of riboflavin transporter deficiency: mini review after five years of experience. *Journal of Inherited Metabolic Disease* 2016. DOI:10.1007/s10545-016-9924-2.

Lake N, Compton A, Rahman S, Thorburn D. Leigh syndrome: One disorder, more than 75 monogenic causes. *Ann Neurol* 2016; **79**: 190–203.

Richards J, Korgenski K, Srivastava R, Bonkowsky J. Costs of the diagnostic odyssey in children with inherited leukodystrophies. *Neurology* 2015; **85**: 1167–70.

Rodan LH, Gibson KM, Pearl PL. Clinical Use of CSF Neurotransmitters. *Pediatr Neurol* 2015;**53**: 277–86.

EPILEPSIE

Allen N, Conroy J, Shahwan A, *et al.*Unexplained early onset epileptic encephalopathy: Exome screening and phenotype expansion. *Epilepsia* 2016; **57**: e12–e17.

Bagnall R, Crompton D, Petrovski S, *et al.* Exome-based analysis of cardiac arrhythmia, respiratory control, and epilepsy genes in sudden unexpected death in epilepsy. *Ann Neurol* 2016; epub.

Chan S, Bello J, Shinnar S, *et al.* Hippocampal Malrotation Is Associated With Prolonged Febrile Seizures: Results of the FEBSTAT Study. *American Journal of Roentgenology* 2015; **205**: 1068–74.

Devinsky O, Marsh E, Friedman D, *et al.* Cannabidiol in patients with treatment-resistant epilepsy: an open-label interventional trial. *The Lancet Neurology* 2016; **15**: 270–8.

Howell, McMahon, Carvill, *et al.* SCN2A encephalopathy: A major cause of epilepsy of infancy with migrating focal seizures. *Neurology* 2015; **85**. DOI:10.1212/WNL.0000000000001926.

Knupp K, Coryell J, Nickels K, *et al.* Response to treatment in a prospective national infantile spasms cohort. *Annals of Neurology* 2016; **79**: 475–84.

Mastrangelo M. Novel Genes of Early-Onset Epileptic Encephalopathies: From Genotype to Phenotypes. *Pediatric Neurology* 2015; **53**: 119–29.

McTague A, Howell K, Cross H, Kurian M, Scheffer I. The genetic landscape of the epileptic encephalopathies of infancy and childhood. *The Lancet Neurology* 2016; **15**: 304–16.

Meglio C, Lesca G, Villeneuve N, *et al.* Epileptic patients with de novo *STXBP1* mutations: Key clinical features based on 24 cases. *Epilepsia* 2015; **56**. DOI:10.1111/epi.13214.

Perucca E. The safety of generic substitution in epilepsy. *The Lancet Neurology* 2016. doi:10.1016/S1474-4422(16)00042-9.

Privitera M, Welty T, Gidal B, *et al.* Generic-to-generic lamotrigine switches in people with epilepsy: the randomised controlled EQUIGEN trial. *The Lancet Neurology* 2016; **15**. doi:10.1016/S1474-4422(16)00014-4.

Shinnar S, Cnaan A, Hu F, *et al.* Long-term outcomes of generalized tonic-clonic seizures in a childhood absence epilepsy trial. *Neurology* 2015;**85**: 1108–14.

Van Schooneveld M, Braun K, van Rijen P, van Nieuwenhuizen O, Jennekens-Schinkel A. The spectrum of long-term cognitive and functional outcome after hemispherectomy in childhood. *European Journal of Paediatric Neurology* 2016. DOI:10.1016/j.ejpn.2016.01.004.

Verbeek NE, van der Maas NA, Sonsma AC, *et al.* Effect of vaccinations on seizure risk and disease course in Dravet syndrome. *Neurology* 2015; published online July 3. DOI:10.1212/WNL.0000000000001855.

ONCOLOGY

Fullerton HJ, Stratton K, Mueller S, *et al.* Recurrent stroke in childhood cancer survivors.*Neurology* 2015; **85**: 1056–64.

Huse JT, Rosenblum MK. The Emerging Molecular Foundations of Pediatric Brain Tumors. *J Child Neurol* 2015; **30**: 1838–50.

Michiels E, Meeteren A, Doz F, Janssens G, van Dalen E. Chemotherapy for children with medulloblastoma. *The Cochrane Library* 2015; **1**: CD006678.

Stucklin A, Tabori U, Grotzer M. The Changing Landscape of Pediatric Low-Grade Gliomas: Clinical Challenges and Emerging Therapies. *Neuropediatrics* 2016; **47**: 070–83.