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Curriculum vitae

Since 2010	Professor (W3), Experimental Neuropediatrics, Charité, Berlin
2008 – 2010	Professor (W2), Developmental Disorders, NeuroCure Clinical Research Center (NCRC), Berlin
2007	Board certification for Neuropediatrics
2003	Assistant professor (C3), Neuropediatric and Genetics Research, Charité, Berlin
2002	Venia legendi (Habilitation) in Pediatrics
1999	Board certification for Pediatrics, Charité, Berlin
1993 – 1996	Residency, Children's Hospital Charité, Berlin
1992 – 1993	Participation in a WHO Polio eradication project at Allahabad, IN
1990 – 1992	Internship: Department of Pediatrics, Uniklinik Köln, Universität zu Köln
1983 – 1989	Studies in Medicine, Universität des Saarlandes; Freie Universität Berlin; University College Dublin, IE; University of Hong Kong, HK

Research fields

Our research program focuses on:

- Neuromuscular and mitochondrial disorders
- Gene hunting for disorders relevant in the field of neuropediatrics and developmental disorders of the nervous system
- Genetic causes of infantile spasms (West syndrome)
- Development of gene finding software and software to handle and interpret next-generation sequencing results
- Muscle growth factors, e.g. myostatin (functional cell biological studies) and satellite cells (muscle regeneration)

Activities in the scientific community, honors, awards

2008 – 2018	Member, editorial board, Journal of Biomedicine and Biotechnology and Academic editor, PLoS ONE
2007	Member, Central Research Commission, Charité, Berlin
2005	Scientific award (First Prize), NeuroWiss e.V. zur Förderung der neurologischen Wissenschaften, Frankfurt
1999 – 2000	First place, clinical teachers ranking, Children's Hospital Charité, Berlin
1997 – 1999	Research fellowship, German Research Foundation (DFG), Nijmegen Center for Mitochondrial Disorders, NL
1984 – 1989	Studienstiftung des deutschen Volkes, German Academic Scholarship Foundation

Selected publications

Lorenz C, Lesimple P, Bukowiecki R, Zink A, Inak G, Mlody B, Singh M, Semtner M, Mah N, Aure K, Leong M, Zabiegalov O, Lyras EM, Pfiffer V, Fauler B, Eichhorst J, Wiesner B, Huebner N, Priller J, Mielke T, Meierhofer D, Izsvak Z, Meier JC, Bouillaud F, Adjaye J, Schuelke M, Wanker EE, Lombes A, Prigione A. Human iPSC-Derived Neural Progenitors Are an Effective Drug Discovery Model for Neurological mtDNA Disorders. Cell Stem Cell 2017; 20:659-674

Knierim E, Hirata H, Wolf NI, Morales-Gonzalez S, Schottmann G, Tanaka Y, Rudnik-Schoneborn S, Orgeur M, Zerres K, Vogt S, van Riesen A, Gill E, Seifert F, Zwirner A, Kirschner J, Goebel HH, Hubner C, Stricker S, Meierhofer D, Stenzel W, Schuelke M. Mutations in Subunits of the Activating Signal Cointegrator 1 Complex Are Associated with Prenatal Spinal Muscular Atrophy and Congenital Bone Fractures. Am J Hum Genet 2016; 98:473-489

Schwarz JM, Cooper DN, Schuelke M, Seelow D. MutationTaster2: mutation prediction for the deep-sequencing age. Nat Methods 2014; 11:361-362

Hirata H, Nanda I, van Riesen A, McMichael G, Hu H, Hambrock M, Papon MA, Fischer U, Marouillat S, Ding C, Alirol S, Bienek M, Preisler-Adams S, Grimme A, Seelow D, Webster R, Haan E, MacLennan A, Stenzel W, Yap TY, Gardner A, Nguyen LS, Shaw M, Lebrun N, Haas SA, Kress W, Haaf T, Schellenberger E, Chelly J, Viot G, Shaffer LG, Rosenfeld JA, Kramer N, Falk R, El-Khechen D, Escobar LF, Hennekam R, Wieacker P, Hubner C, Ropers HH, Gecz J, Schuelke M*, Laumonnier F, Kalscheuer VM*. ZC4H2 mutations are associated with arthrogryposis multiplex congenita and intellectual disability through impairment of central and peripheral synaptic plasticity. Am J Hum Genet 2013; 92:681-695 | *corresponding authors

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, Hubner 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; 43:1189-1192 | *corresponding authors

Schwarz JM, Rodelsperger C, Schuelke M, Seelow D. MutationTaster evaluates disease-causing potential of sequence alterations. Nat Methods 2010; 7:575-576

Schuelke M, Wagner KR, Stolz LE, Hubner C, Riebel T, Komen W, Braun T, Tobin JF, Lee SJ. Myostatin mutation associated with gross muscle hypertrophy in a child. N Engl J Med 2004; 350:2682-2688

Grohmann K, Schuelke M, Diers A, Hoffmann K, Lucke B, Adams C, Bertini E, Leonhardt-Horti H, Muntoni F, Ouvrier R, Pfeufer A, Rossi R, Van Maldergem L, Wilmshurst JM, Wienker TF, Sendtner M, Rudnik-Schoneborn S, Zerres K, Hubner C. Mutations in the gene encoding immunoglobulin mu-binding protein 2 cause spinal muscular atrophy with respiratory distress type 1. Nat Genet 2001; 29:75-77

Schuelke M. An economic method for the fluorescent labeling of PCR fragments. Nat Biotechnol 2000; 18:233-234

Schuelke M, Smeitink J, Mariman E, Loeffen J, Plecko B, Trijbels F, Stockler-Ipsiroglu S, van den Heuvel L. Mutant NDUFV1 subunit of mitochondrial complex I causes leukodystrophy and myoclonic epilepsy. Nat Genet 1999; 21:260-261