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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
1997 – 1999	DFG Research Fellowship, Nijmegen Center for Mitochondrial Disorders, NL
1993 – 1996	Residency, Children's Hospital, Charité, Berlin
1993	Doctoral degree (Dr. med.), Institute of Neuropathology, Freie Universität Berlin
1992 – 1993	Participation in a WHO Polio eradication project at Allahabad, IN
1990 – 1992	Internship: Department of Pediatrics, 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:

- Gene finding projects for disorders relevant in the field of neuropediatrics and developmental disorders of the nervous system
- Development of gene finding software and of software to handle and interpret NGS results.
- *Arthrogryposis multiplex congenita*
- Development of the neuromuscular unit
- Transcription factor biology
- Repurposing of drugs using human iPSC models

Activities in the scientific community, honors, awards

Since 2019	Speaker, DFG Research Unit FOR2841 'Beyond the exome'
Since 2017	Member, scientific board, 'Eva Luise und Horst Köhler Stiftung für Menschen mit Seltenen Erkrankungen'
Since 2008	Principal investigator, NeuroCure – Cluster of Excellence, Berlin
Since 2008	Academic editor, PLoS ONE
2008 – 2018	Member, editorial board, Journal of Biomedicine and Biotechnology
Since 2007	Member, Charité Ethics Committee (Deputy Chairman since 2010), 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	Scholarship, Studienstiftung des deutschen Volkes (German Academic Scholarship Foundation)

Selected publications

Gusic M, Schottmann G, Feichtinger RG, Du C, Scholz C, Wagner M, Mayr JA, Lee CY, Yepez VA, Lorenz N, Morales-Gonzalez S, Panneman DM, Rotig A, Rodenburg RJT, Wortmann SB, Prokisch H, Schuelke M. Bi-Allelic UQCRFS1 Variants Are Associated with Mitochondrial Complex III Deficiency, Cardiomyopathy, and Alopecia Totalis. Am J Hum Genet 2020; 106: 102-111

Mueller N, Sassa T, Morales-Gonzalez S, Schneider J, Salchow DJ, Seelow D, Knierim E, Stenzel W, Kihara A, Schuelke M. De novo mutation in ELOVL1 causes ichthyosis, acanthosis nigricans, hypomyelination, spastic paraplegia, high frequency deafness and optic atrophy. J Med Genet 2019; 56: 164-175

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

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

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