

## Markus Schülke-Gerstenfeld

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### Curriculum vitae

since 2010	Professorship (W3, tenured), Experimental Neuropediatrics, Charité
2008 – 2010	Professor (W2), Department Director, Developmental Disorders, NeuroCure Clinical Research Center
2007	Board certification for Neuropediatrics
2003	Assistant Professor (C3), Neuropediatric and genetics research, Charité
2002	Faculty rank (Habilitation) in Pediatrics
1999	Board certification for Pediatrics
1993 – 1996	Residency: University Children's Hospital, Charité
1992 – 1993	Participation in a WHO Polio eradication project at Allahabad, U.P. (India)
1990 – 1992	Internship: University Children's Hospital, University of Cologne
1983 – 1989	Medical School: University of the Saarland (Homburg), Freie Universität Berlin, University College Dublin, University of Hong Kong

### Research fields

- 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 – 2013	Member, Editorial Board, Journal of Biomedicine and Biotechnology and Academic Editor, PLoS ONE
2007	Member, central research commission, Charité
2005	Scientific award (First Prize), NeuroWiss Verein zur Förderung der Neurologischen Wissenschaften, Frankfurt
1999 – 2000	First place, clinical teachers ranking, Children's Hospital Charité
1997 – 1999	Research fellowship, German Research Foundation (DFG), Nijmegen Center for Mitochondrial Disorders, The Netherlands
1984 – 1989	„Studienstiftung des deutschen Volkes“

## Selected publications

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(3):473-89.

Schwarz JM, Cooper DN, Schuelke M, Seelow D. MutationTaster2: mutation prediction for the deep-sequencing age. *Nat Methods.* 2014;11(4):361-2.

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(5):681-95.

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(12):1189-92.

Kann O, Huchzermeyer C, Kovacs R, Wirtz S, Schuelke M. Gamma oscillations in the hippocampus require high complex I gene expression and strong functional performance of mitochondria. *Brain.* 2011;134(Pt 2):345-58. doi: 10.1093/brain/awq333. PubMed PMID: 21183487.

Schwarz JM, Rodelsperger C, Schuelke M, Seelow D. MutationTaster evaluates disease-causing potential of sequence alterations. *Nat Methods.* 2010;7(8):575-6.

Rajab A, Straub V, McCann LJ, Seelow D, Varon R, Barresi R, Schulze A, Lucke B, Lutzkendorf S, Karbasiyan M, Bachmann S, Spuler S, Schuelke M. Fatal cardiac arrhythmia and long-QT syndrome in a new form of congenital generalized lipodystrophy with muscle rippling (CGL4) due to PTRF-CAVIN mutations. *PLoS Genet.* 2010;6(3):e1000874.

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(26):2682-8.

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(1):75-7.

Schuelke M. An economic method for the fluorescent labeling of PCR fragments. *Nat Biotechnol.* 2000;18(2):233-4.

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(3):260-1.