

Prof. Dr. med. Markus Schülke-Gerstenfeld
(* 1962)



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Curriculum vitae

- since 2003 C3 Professorship for Neuropediatric and Genetics Research at the Charité – Universitätsmedizin Berlin
- since 2002 Habilitation in Pediatrics with the subject: ‘Clinical, biochemical and molecular studies in children with mitochondrial diseases’
- since 1999 Consultant Neuropediatrician
- 1999 Board certification for Pediatrics
- 1997 - 1999 Research scholarship of the German Research Foundation (DFG) at the “Nijmegen Center for Mitochondrial Disorders” in the Netherlands
- 1993 - 1996 Resident at the Department of Neuropediatrics at the Freie Universität, Berlin and subsequently at the Charité
- 1992 - 1993 WHO Polio eradication project at Allahabad (India)
- 1992 PhD thesis at the Institute of Neuropathology, Freie Universität, Berlin
- 1990 - 1992 Internship at the University Childrens’ Hospital Cologne
- 1983 - 1989 Studies in medicine in Homburg (Saar), Berlin, Dublin and Hong Kong

Research fields

Our group is active in the field of genetics research, biochemistry and cell biology with the following key areas:

- Mitochondrial diseases
- Neuromuscular diseases
- Ataxia with isolated Vitamin E deficiency
- Essential tremor

Activities in the scientific community, honors, awards

- 2005 Scientific Award, NeuroWiss, Association for the Promotion of Neurological Sciences (Verein zur Förderung der Neurologischen Wissenschaften), Frankfurt
- 1999 - 2000 First place in ranking of clinical teachers at the Children’s Hospital of the Charité
- 1997 - 1999 Research Scholarship, Nijmegen Center for Mitochondrial Disorders, The Netherlands (DFG)
- 1984 - 1989 Scholarship, German National Merit Foundation (Studienstiftung des Deutschen Volkes)

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Selected publications

Guenther U, Varon R, Schlicke M, Dutrannoy V, Volk A, Hubner C, von Au K Schuelke M. (2007) Clinical and mutational profile in spinal muscular atrophy with respiratory distress (SMARD). Defining novel phenotypes through Hierarchical Cluster Analysis. *Hum Mutat* – in press

Brinckmann A, Ruther K, Williamson K, Lorenz B, Lucke B, Nurnberg P, Trijbels F, Janssen A, Schuelke M. (2007) De novo double mutation in PAX6 and mtDNA tRNA(Lys) associated with atypical aniridia and mitochondrial disease. *J Mol Med* 85:163-8

Lopez LC, Schuelke M, Quinzii CM, Kanki T, Rodenburg RJ, Naini A, Dimauro S, Hirano M. (2006) Leigh syndrome with nephropathy and CoQ10 deficiency due to decaprenyl diphosphate synthase subunit 2 (PDSS2) mutations. *Am J Hum Genet* 79:1125-9

Xie J, Techritz S, Haebel S, Horn A, Neitzel H, Klose J, Schuelke M (2005) A two-dimensional electrophoretic map of human mitochondrial proteins from lymphoblastoid cell lines. A prerequisite to study mitochondrial disorders in patients. *Proteomics* 5:2981-99

Schuelke M, Wagner KR, Stolz LE, Hubner C, Riebel T, Komen W, Braun T, Tobin JF, Lee SJ (2004) Myostatin mutation associated with gross muscle hypertrophy in a child. *N Engl J Med* 350:2682-8 (author's reply to Correspondance: Uhlenberg B, Lucke B, Schuelke M (2004) *N Engl J Med* 352:1030-1)

Uhlenberg B, Schuelke M, Ruschendorf F, Ruf N, Kaindl AM, Henneke M, Thiele H, Stoltenburg-Didinger G, Aksu F, Topaloglu H, Nürnberg P, Hübner C, Weschke B, Gärtner J (2004) Mutations in the gene encoding gap junction protein alpha 12 (Connexin 46.6) cause Pelizaeus-Merzbacher-like disease. *Am J Hum Genet* 75:251-60

Schuelke M, Krude H, Finckh B, Mayatepek E, Janssen A, Schmelz M, Trefz F, Trijbels F, Smeitink J (2002) Septo–optic dysplasia associated with a new mitochondrial cytochrome b–mutation. *Ann Neurol* 51:388-92

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–Schöneborn S, Zerres K, Hübner C (2001) Mutations in the gene encoding immunoglobulin–binding protein 2 cause spinal muscular atrophy with respiratory distress type 1. *Nat Genet* 29:75-7

Schuelke M (2000) An economic method for fluorescent labeling of PCR fragments. A poor man's approach to genotyping. *Nat Biotechnol* 18:233-4

Schuelke M, Smeitink J, Mariman E, Loeffen J, Plecko B, Trijbels F, Stockler-Ipsiroglu S, van den Heuvel L (1999) Mutant NDUFV1-subunit of mitochondrial complex I causes leukodystrophy and myoclonic epilepsy. *Nat Genet* 21: 260-1