Markus Schuelke

List of Publications by Citations

171 papers	15,530 citations	48 h-index	124 g-index
185 ext. papers	17,743 ext. citations	8.2 avg, IF	6.75 L-index

#	Paper	IF	Citations
171	An economic method for the fluorescent labeling of PCR fragments. *Nature Biotechnology*, **2000**, 18, 233-4	44.5	2709
170	MutationTaster2: mutation prediction for the deep-sequencing age. *Nature Methods*, **2014**, 11, 361-2	21.6	2455
169	MutationTaster evaluates disease-causing potential of sequence alterations. *Nature Methods*, **2010**, 7, 575-6	21.6	2091
168	Myostatin mutation associated with gross muscle hypertrophy in a child. *New England Journal of Medicine*, **2004**, 350, 2682-8	59.2	1044
167	Leigh syndrome with nephropathy and CoQ10 deficiency due to decaprenyl diphosphate synthase subunit 2 (PDSS2) mutations. *American Journal of Human Genetics*, **2006**, 79, 1125-9	11	324
166	Lack of myostatin results in excessive muscle growth but impaired force generation. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 1835-40	11.5	293
165	HomozygosityMapper--an interactive approach to homozygosity mapping. *Nucleic Acids Research*, **2009**, 37, W593-9	20.1	289
164	Mutations in the gene encoding immunoglobulin mu-binding protein 2 cause spinal muscular atrophy with respiratory distress type 1. *Nature Genetics*, **2001**, 29, 75-7	36.3	272
163	Mutant NDUFV1 subunit of mitochondrial complex I causes leukodystrophy and myoclonic epilepsy. *Nature Genetics*, **1999**, 21, 260-1	36.3	238
162	The first nuclear-encoded complex I mutation in a patient with Leigh syndrome. *American Journal of Human Genetics*, **1998**, 63, 1598-608	11	232
161	Mutations in the gene encoding gap junction protein alpha 12 (connexin 46.6) cause Pelizaeus-Merzbacher-like disease. *American Journal of Human Genetics*, **2004**, 75, 251-60	11	227
160	Variants in CPA1 are strongly associated with early onset chronic pancreatitis. *Nature Genetics*, **2013**, 45, 1216-20	36.3	192
159	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 Genetics*, **2010**, 6, e1000874	6	178
158	Mutations in PYCR1 cause cutis laxa with progeroid features. *Nature Genetics*, **2009**, 41, 1016-21	36.3	178
157	The spectrum of WRN mutations in Werner syndrome patients. *Human Mutation*, **2006**, 27, 558-67	4.7	172
156	Genotypic and phenotypic spectrum of PANK2 mutations in patients with neurodegeneration with brain iron accumulation. *Annals of Neurology*, **2006**, 59, 248-56	9.4	158
155	Molecular diagnosis in mitochondrial complex I deficiency using exome sequencing. *Journal of Medical Genetics*, **2012**, 49, 277-83	5.8	145

154	Identification of small non-coding RNAs from mitochondria and chloroplasts. *Nucleic Acids Research* , **2006**, 34, 3842-52	20.1	144
153	Gamma oscillations in the hippocampus require high complex I gene expression and strong functional performance of mitochondria. *Brain*, **2011**, 134, 345-58	11.2	121
152	Reactive oxygen species, oxidative stress, and cell death correlate with level of CoQ10 deficiency. *FASEB Journal*, **2010**, 24, 3733-43	0.9	117
151	Respiratory chain dysfunction and oxidative stress correlate with severity of primary CoQ10 deficiency. *FASEB Journal*, **2008**, 22, 1874-85	0.9	114
150	Selective disactivation of neurofibromin GAP activity in neurofibromatosis type 1. *Human Molecular Genetics*, **1998**, 7, 1261-8	5.6	113
149	Infantile spinal muscular atrophy with respiratory distress type 1 (SMARD1). *Annals of Neurology*, **2003**, 54, 719-24	9.4	112
148	Synaptic PRG-1 modulates excitatory transmission via lipid phosphate-mediated signaling. *Cell*, **2009**, 138, 1222-35	56.2	100
147	GeneDistiller--distilling candidate genes from linkage intervals. *PLoS ONE*, **2008**, 3, e3874	3.7	88
146	Systematic comparison of three methods for fragmentation of long-range PCR products for next generation sequencing. *PLoS ONE*, **2011**, 6, e28240	3.7	87
145	Dynamics of myosin degradation in intensive care unit-acquired weakness during severe critical illness. *Intensive Care Medicine*, **2014**, 40, 528-38	14.5	85
144	Human iPSC-Derived Neural Progenitors Are an Effective Drug Discovery Model for Neurological mtDNA Disorders. *Cell Stem Cell*, **2017**, 20, 659-674.e9	18	84
143	Treatment of CoQ(10) deficient fibroblasts with ubiquinone, CoQ analogs, and vitamin C: time- and compound-dependent effects. *PLoS ONE*, **2010**, 5, e11897	3.7	82
142	EXOSC8 mutations alter mRNA metabolism and cause hypomyelination with spinal muscular atrophy and cerebellar hypoplasia. *Nature Communications*, **2014**, 5, 4287	17.4	80
141	Mutations in MEGF10, a regulator of satellite cell myogenesis, cause early onset myopathy, areflexia, respiratory distress and dysphagia (EMARDD). *Nature Genetics*, **2011**, 43, 1189-92	36.3	71
140	Deficiency of ECHS1 causes mitochondrial encephalopathy with cardiac involvement. *Annals of Clinical and Translational Neurology*, **2015**, 2, 492-509	5.3	69
139	IGHMBP2 is a ribosome-associated helicase inactive in the neuromuscular disorder distal SMA type 1 (DSMA1). *Human Molecular Genetics*, **2009**, 18, 1288-300	5.6	69
138	Septo-optic dysplasia associated with a new mitochondrial cytochrome b mutation. *Annals of Neurology*, **2002**, 51, 388-92	9.4	69
137	cDNA of eight nuclear encoded subunits of NADH:ubiquinone oxidoreductase: human complex I cDNA characterization completed. *Biochemical and Biophysical Research Communications*, **1998**, 253, 415-22	3.4	63

136	TRMT5 Mutations Cause a Defect in Post-transcriptional Modification of Mitochondrial tRNA Associated with Multiple Respiratory-Chain Deficiencies. *American Journal of Human Genetics*, **2015**, 97, 319-28	11	62
135	Epilepsia partialis continua associated with a homoplasmic mitochondrial tRNA(Ser(UCN)) mutation. *Annals of Neurology*, **1998**, 44, 700-4	9.4	61
134	Comprehensive genotyping and clinical characterisation reveal 27 novel NKX2-1 mutations and expand the phenotypic spectrum. *Journal of Medical Genetics*, **2014**, 51, 375-87	5.8	60
133	Treatment of ataxia in isolated vitamin E deficiency caused by alpha-tocopherol transfer protein deficiency. *Journal of Pediatrics*, **1999**, 134, 240-4	3.6	59
132	Urinary □tocopherol metabolites in □tocopherol transfer protein-deficient patients. *Journal of Lipid Research*, **2000**, 41, 1543-1551	6.3	59
131	Phenotypic spectrum of eleven patients and five novel MTFMT mutations identified by exome sequencing and candidate gene screening. *Molecular Genetics and Metabolism*, **2014**, 111, 342-352	3.7	58
130	Clinical and mutational profile in spinal muscular atrophy with respiratory distress (SMARD): defining novel phenotypes through hierarchical cluster analysis. *Human Mutation*, **2007**, 28, 808-15	4.7	55
129	Blockade of ActRIIB signaling triggers muscle fatigability and metabolic myopathy. *Molecular Therapy*, **2014**, 22, 1423-1433	11.7	54
128	ZC4H2 mutations are associated with arthrogryposis multiplex congenita and intellectual disability through impairment of central and peripheral synaptic plasticity. *American Journal of Human Genetics*, **2013**, 92, 681-95	11	54
127	Urinary alpha-tocopherol metabolites in alpha-tocopherol transfer protein-deficient patients. *Journal of Lipid Research*, **2000**, 41, 1543-51	6.3	54
126	HomozygosityMapper2012--bridging the gap between homozygosity mapping and deep sequencing. *Nucleic Acids Research*, **2012**, 40, W516-20	20.1	52
125	Myostatin is a key mediator between energy metabolism and endurance capacity of skeletal muscle. *American Journal of Physiology - Regulatory Integrative and Comparative Physiology*, **2014**, 307, R444-54	3.2	50
124	Comparative analysis of uncoupling protein 4 distribution in various tissues under physiological conditions and during development. *Biochimica Et Biophysica Acta - Biomembranes*, **2009**, 1788, 2309-19	3.8	50
123	CoQ deficiency causes disruption of mitochondrial sulfide oxidation, a new pathomechanism associated with this syndrome. *EMBO Molecular Medicine*, **2017**, 9, 78-95	12	47
122	KIF1C mutations in two families with hereditary spastic paraparesis and cerebellar dysfunction. *Journal of Medical Genetics*, **2014**, 51, 137-42	5.8	46
121	Spinal muscular atrophy with respiratory distress type 1 (SMARD1). *Journal of Child Neurology*, **2008**, 23, 199-204	2.5	46
120	Heterozygous myogenic factor 6 mutation associated with myopathy and severe course of Becker muscular dystrophy. *Neuromuscular Disorders*, **2000**, 10, 572-7	2.9	45
119	Coenzyme Q deficiency causes impairment of the sulfide oxidation pathway. *EMBO Molecular Medicine*, **2017**, 9, 96-111	12	44

118	NOA1 is an essential GTPase required for mitochondrial protein synthesis. *Molecular Biology of the Cell*, **2011**, 22, 1-11	3.5	44
117	The natural course of infantile spinal muscular atrophy with respiratory distress type 1 (SMARD1). *Pediatrics*, **2012**, 129, e148-56	7.4	43
116	Mutations in Subunits of the Activating Signal Cointegrator 1 Complex Are Associated with Prenatal Spinal Muscular Atrophy and Congenital Bone Fractures. *American Journal of Human Genetics*, **2016**, 98, 473-489	11	42
115	International Workshop:: Outcome measures and clinical trial readiness in primary mitochondrial myopathies in children and adults. Consensus recommendations. 16-18 November 2016, Rome, Italy. *Neuromuscular Disorders*, **2017**, 27, 1126-1137	2.9	42
114	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. *Orphanet Journal of Rare Diseases*, **2018**, 13, 120	4.2	41
113	Potassium channel KIR4.1-specific antibodies in children with acquired demyelinating CNS disease. *Neurology*, **2014**, 82, 470-3	6.5	41
112	POMK mutation in a family with congenital muscular dystrophy with merosin deficiency, hypomyelination, mild hearing deficit and intellectual disability. *Journal of Medical Genetics*, **2014**, 51, 275-82	5.8	40
111	Localization of alpha-tocopherol transfer protein in trophoblast, fetal capillaries' endothelium and amnion epithelium of human term placenta. *Free Radical Research*, **2004**, 38, 413-20	4	40
110	Two patients with MIRAGE syndrome lacking haematological features: role of somatic second-site reversion SAMD9 mutations. *Journal of Medical Genetics*, **2018**, 55, 81-85	5.8	38
109	De Novo Mutations in SLC25A24 Cause a Craniosynostosis Syndrome with Hypertrichosis, Progeroid Appearance, and Mitochondrial Dysfunction. *American Journal of Human Genetics*, **2017**, 101, 833-843	11	37
108	Variants in EXOSC9 Disrupt the RNA Exosome and Result in Cerebellar Atrophy with Spinal Motor Neuronopathy. *American Journal of Human Genetics*, **2018**, 102, 858-873	11	37
107	Mammalian mitochondrial nitric oxide synthase: characterization of a novel candidate. *FEBS Letters*, **2006**, 580, 455-62	3.8	37
106	Transparent Danionella translucida as a genetically tractable vertebrate brain model. *Nature Methods*, **2018**, 15, 977-983	21.6	37
105	Recurrent stroke due to a novel voltage sensor mutation in Cav2.1 responds to verapamil. *Stroke*, **2011**, 42, e14-7	6.7	36
104	De novo mutation in causes ichthyosis, , hypomyelination, spastic paraplegia, high frequency deafness and optic atrophy. *Journal of Medical Genetics*, **2019**, 56, 164-175	5.8	35
103	A novel mutation in two Turkish families associated with cerebral atrophy, global retardation, scoliosis, achalasia and alacrima. *Journal of Medical Genetics*, **2017**, 54, 176-185	5.8	34
102	Increased mRNA expression of tissue inhibitors of metalloproteinase-1 and -2 in Duchenne muscular dystrophy. *Acta Neuropathologica*, **2005**, 109, 285-93	14.3	34
101	A recessive mutation in beta-IV-spectrin (SPTBN4) associates with congenital myopathy, neuropathy, and central deafness. *Human Genetics*, **2017**, 136, 903-910	6.3	32

100	Clinical variability in distal spinal muscular atrophy type 1 (DSMA1): determination of steady-state IGHMBP2 protein levels in five patients with infantile and juvenile disease. *Journal of Molecular Medicine*, **2009**, 87, 31-41	5.5	31
99	Neonatal lactic acidosis, complex I/IV deficiency, and fetal cerebral disruption. *Neuropediatrics*, **2005**, 36, 193-9	1.6	31
98	Spinal muscular atrophy-like picture, cardiomyopathy, and cytochrome c oxidase deficiency. *Neurology*, **1999**, 52, 383-6	6.5	31
97	Genomic rearrangements at the IGHMBP2 gene locus in two patients with SMARD1. *Human Genetics*, **2004**, 115, 319-26	6.3	28
96	Nemaline body myopathy caused by a novel mutation in troponin T1 (TNNT1). *Muscle and Nerve*, **2016**, 53, 564-9	3.4	28
95	Liver transplantation: treatment of choice for hepatic and neurological manifestation of Wilson's disease. *Clinical Transplantation*, **1997**, 11, 217-24	3.8	28
94	Altered RNA metabolism due to a homozygous RBM7 mutation in a patient with spinal motor neuropathy. *Human Molecular Genetics*, **2016**, 25, 2985-2996	5.6	27
93	BMP signaling regulates satellite cell-dependent postnatal muscle growth. *Development (Cambridge)*, **2017**, 144, 2737-2747	6.6	25
92	Combined effect of AAV-U7-induced dystrophin exon skipping and soluble activin Type IIB receptor in mdx mice. *Human Gene Therapy*, **2012**, 23, 1269-79	4.8	25
91	Muscle and nerve pathology in Dunnigan familial partial lipodystrophy. *Neurology*, **2007**, 68, 677-83	6.5	24
90	Analysis of mitochondrial DNA in discordant monozygotic twins with neurofibromatosis type 1. *Twin Research and Human Genetics*, **2007**, 10, 486-95	2.2	24
89	Muscle 3243A-->G mutation load and capacity of the mitochondrial energy-generating system. *Annals of Neurology*, **2008**, 63, 473-81	9.4	23
88	MutationDistiller: user-driven identification of pathogenic DNA variants. *Nucleic Acids Research*, **2019**, 47, W114-W120	20.1	22
87	A two-dimensional electrophoretic map of human mitochondrial proteins from immortalized lymphoblastoid cell lines: a prerequisite to study mitochondrial disorders in patients. *Proteomics*, **2005**, 5, 2981-99	4.8	22
86	A movement disorder with dystonia and ataxia caused by a mutation in the HIBCH gene. *Movement Disorders*, **2016**, 31, 1733-1739	7	21
85	Protracted course of juvenile ceroid lipofuscinosis associated with a novel CLN3 mutation (p.Y199X). *Clinical Genetics*, **2009**, 76, 38-45	4	21
84	Human muscle-derived CLEC14A-positive cells regenerate muscle independent of PAX7. *Nature Communications*, **2019**, 10, 5776	17.4	21
83	MORC2 mutation causes severe spinal muscular atrophy-phenotype, cerebellar atrophy, and diaphragmatic paralysis. *Brain*, **2016**, 139, e70	11.2	20

82	Region-specific expression of mitochondrial complex I genes during murine brain development. *PLoS ONE*, **2011**, 6, e18897	3.7	20
81	Regionalized pathology correlates with augmentation of mtDNA copy numbers in a patient with myoclonic epilepsy with ragged-red fibers (MERRF-syndrome). *PLoS ONE*, **2010**, 5, e13513	3.7	19
80	Cloning of the human mitochondrial 51 kDa subunit (NDUFV1) reveals a 100% antisense homology of its 3'UTR with the 5'UTR of the gamma-interferon inducible protein (IP-30) precursor: is this a link between mitochondrial myopathy and inflammation?. *Biochemical and Biophysical Research Communications*, **1998**, 245, 599-606	3.4	19
79	Bi-Allelic UQCRFS1 Variants Are Associated with Mitochondrial Complex III Deficiency, Cardiomyopathy, and Alopecia Totalis. *American Journal of Human Genetics*, **2020**, 106, 102-111	11	19
78	Recessive DEAF1 mutation associates with autism, intellectual disability, basal ganglia dysfunction and epilepsy. *Journal of Medical Genetics*, **2015**, 52, 607-11	5.8	18
77	BRAT1 mutations are associated with infantile epileptic encephalopathy, mitochondrial dysfunction, and survival into childhood. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2274-81	2.5	18
76	Recessive mutation in EXOSC3 associates with mitochondrial dysfunction and pontocerebellar hypoplasia. *Mitochondrion*, **2017**, 37, 46-54	4.9	17
75	Multiple origins of the mtDNA 7472insC mutation associated with hearing loss and neurological dysfunction. *European Journal of Human Genetics*, **2001**, 9, 385-7	5.3	17
74	Defective metabolic programming impairs early neuronal morphogenesis in neural cultures and an organoid model of Leigh syndrome. *Nature Communications*, **2021**, 12, 1929	17.4	17
73	Hybrid genome assembly and annotation of Danionella translucida. *Scientific Data*, **2019**, 6, 156	8.2	16
72	Recessive REEP1 mutation is associated with congenital axonal neuropathy and diaphragmatic palsy. *Neurology: Genetics*, **2015**, 1, e32	3.8	16
71	Clinical application of whole exome sequencing reveals a novel compound heterozygous TK2-mutation in two brothers with rapidly progressive combined muscle-brain atrophy, axonal neuropathy, and status epilepticus. *Mitochondrion*, **2015**, 20, 1-6	4.9	15
70	Recessive truncating IGHMBP2 mutations presenting as axonal sensorimotor neuropathy. *Neurology*, **2015**, 84, 523-31	6.5	15
69	New Nuclear Encoded Mitochondrial Mutation Illustrates Pitfalls in Prenatal Diagnosis by Biochemical Methods. *Clinical Chemistry*, **2002**, 48, 772-775	5.5	15
68	Morvan syndrome associated with CASPR2 and LGI1 antibodies in a child. *Neurology*, **2018**, 90, 183-185	6.5	14
67	Characterization of a Dmd (EGFP) reporter mouse as a tool to investigate dystrophin expression. *Skeletal Muscle*, **2016**, 6, 25	5.1	14
66	MutationTaster2021. *Nucleic Acids Research*, **2021**, 49, W446-W451	20.1	14
65	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. *Brain*, **2019**, 142, 2948-2964	11.2	13

64	New evidence for a mutation hotspot in exon 37 of the NF1 gene. *Human Mutation*, **1997**, 9, 374-7	4.7	12
63	Improved glucose metabolism in mice lacking alpha-tocopherol transfer protein. *European Journal of Nutrition*, **2007**, 46, 397-405	5.2	12
62	Ataxia with vitamin E deficiency: biochemical effects of malcompliance with vitamin E therapy. *Neurology*, **2000**, 55, 1584-6	6.5	12
61	CARbon DIoxide for the treatment of Febrile seizures: rationale, feasibility, and design of the CARDIF-study. *Journal of Translational Medicine*, **2013**, 11, 157	8.5	11
60	Kyphoscoliosis peptidase (KY) mutation causes a novel congenital myopathy with core targetoid defects. *Acta Neuropathologica*, **2016**, 132, 475-8	14.3	11
59	Myopathology in the times of modern genetics. *Neuropathology and Applied Neurobiology*, **2017**, 43, 44-61	5.2	10
58	A systematic, large-scale comparison of transcription factor binding site models. *BMC Genomics*, **2016**, 17, 388	4.5	10
57	RegulationSpotter: annotation and interpretation of extratranscriptic DNA variants. *Nucleic Acids Research*, **2019**, 47, W106-W113	20.1	9
56	A novel frameshift mutation of C19ORF12 causes NBIA4 with cerebellar atrophy and manifests with severe peripheral motor axonal neuropathy. *Clinical Genetics*, **2014**, 85, 290-2	4	9
55	De novo double mutation in PAX6 and mtDNA tRNA(Lys) associated with atypical aniridia and mitochondrial disease. *Journal of Molecular Medicine*, **2007**, 85, 163-8	5.5	9
54	Complement deposition at the neuromuscular junction in seronegative myasthenia gravis. *Acta Neuropathologica*, **2020**, 139, 1119-1122	14.3	8
53	Infant botulism: is there an association with thiamine deficiency?. *Pediatrics*, **2014**, 134, e1436-40	7.4	8
52	Familial glucocorticoid deficiency type 1 due to a novel compound heterozygous MC2R mutation. *Hormone Research in Paediatrics*, **2008**, 69, 363-8	3.3	8
51	Detection of novel NF1 mutations and rapid mutation prescreening with Pyrosequencing. *Electrophoresis*, **2007**, 28, 4295-301	3.6	8
50	Quantitative and qualitative 2D electrophoretic analysis of differentially expressed mitochondrial proteins from five mouse organs. *Proteomics*, **2013**, 13, 179-95	4.8	7
49	Identifying dynamic membrane structures with atomic-force microscopy and confocal imaging. *Microscopy and Microanalysis*, **2014**, 20, 514-20	0.5	7
48	Tandem duplication of DMD exon 18 associated with epilepsy, macroglossia, and endocrinologic abnormalities. *Muscle and Nerve*, **2007**, 35, 396-401	3.4	7
47	A novel homozygous nonsense mutation of VPS13B associated with previously unreported features of Cohen syndrome. *American Journal of Medical Genetics, Part A*, **2020**, 182, 570-575	2.5	7

46	A homozygous PIGO mutation associated with severe infantile epileptic encephalopathy and corpus callosum hypoplasia, but normal alkaline phosphatase levels. *Metabolic Brain Disease*, **2017**, 32, 2131-2137	3.9	6
45	Caveolin 1 Promotes Renal Water and Salt Reabsorption. *Scientific Reports*, **2018**, 8, 545	4.9	6
44	Extracellular matrix remodelling is associated with muscle force increase in overloaded mouse plantaris muscle. *Neuropathology and Applied Neurobiology*, **2021**, 47, 218-235	5.2	6
43	Autophagic vacuolar myopathy is a common feature of CLN3 disease. *Annals of Clinical and Translational Neurology*, **2018**, 5, 1385-1393	5.3	6
42	Leukodystrophy with multiple beaded periventricular cysts: unusual cranial MRI results in Canavan disease. *Journal of Inherited Metabolic Disease*, **2015**, 38, 983-4	5.4	5
41	Muscle Weakness, Cardiomyopathy, and L-2-Hydroxyglutaric Aciduria Associated with a Novel Recessive SLC25A4 Mutation. *JIMD Reports*, **2019**, 43, 27-35	1.9	5
40	A new mutation of IGHMBP2 gene. *Pediatric Neurology*, **2006**, 34, 168	2.9	5
39	Motor function in survivors of pediatric acute lymphoblastic leukemia treated with chemotherapy-only. *European Journal of Paediatric Neurology*, **2019**, 23, 304-316	3.8	5
38	Allan-Herndon-Dudley-Syndrome: Considerations about the Brain Phenotype with Implications for Treatment Strategies. *Experimental and Clinical Endocrinology and Diabetes*, **2020**, 128, 414-422	2.3	4
37	Live-imaging of revertant and therapeutically restored dystrophin in the Dmd mouse model for Duchenne muscular dystrophy. *Neuropathology and Applied Neurobiology*, **2020**, 46, 602-614	5.2	3
36	Kl□er-Bucy syndrome associated with a recessive variant in HGSNAT in two siblings with Mucopolysaccharidosis type IIIC (Sanfilippo C). *European Journal of Human Genetics*, **2017**, 25, 253-256	5.3	3
35	CNVinspector: a web-based tool for the interactive evaluation of copy number variations in single patients and in cohorts. *Journal of Medical Genetics*, **2013**, 50, 529-33	5.8	3
34	Degenerative changes in unmyelinated nerve fibers in late-infantile neuronal ceroidlipofuscinosis. A morphometric study of conjunctival biopsy specimens. *Acta Neuropathologica*, **1998**, 95, 175-83	14.3	3
33	Prenatal manifestation of pancytopenia in Pearson marrow-pancreas syndrome caused by a mitochondrial DNA deletion. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 285-8	2.5	3
32	Genetic deafness in a preterm infant with a critical postnatal course. *Pediatric Critical Care Medicine*, **2006**, 7, 270-2	3	3
31	Defining the ATPome reveals cross-optimization of metabolic pathways. *Nature Communications*, **2020**, 11, 4319	17.4	3
30	Diagnosing pediatric mitochondrial disease: lessons from 2,000 exomes		3
29	Phenotero: Annotate as you write. *Clinical Genetics*, **2019**, 95, 287-292	4	3

28	Cavin 1 function does not follow caveolar morphology. *American Journal of Physiology - Cell Physiology*, **2015**, 308, C1023-30	5.4	2
27	A spontaneous missense mutation in the chromodomain helicase DNA-binding protein 8 (CHD8) gene: a novel association with congenital myasthenic syndrome. *Neuropathology and Applied Neurobiology*, **2020**, 46, 588-601	5.2	2
26	Das Deutsche Netzwerk f mitochondriale Erkrankungen (mitoNET). *Medizinische Genetik*, **2012**, 24, 193-199	0.5	2
25	Hybrid genome assembly and annotation of Danionella translucida, a transparent fish with the smallest known vertebrate brain		2
24	A new homozygous HERC1 gain-of-function variant in MDFPMR syndrome leads to mTORC1 hyperactivation and reduced autophagy during cell catabolism. *Molecular Genetics and Metabolism*, **2020**, 131, 126-134	3.7	2
23	Inflammation, fibrosis and skeletal muscle regeneration in LGMDR9 are orchestrated by macrophages. *Neuropathology and Applied Neurobiology*, **2021**, 47, 856-866	5.2	2
22	Expanding the clinical and molecular spectrum of ATP6V1A related metabolic cutis laxa. *Journal of Inherited Metabolic Disease*, **2021**, 44, 972-986	5.4	2
21	Fulminant cerebral venous thrombosis associated with the m.3243A>G MELAS mutation: A new guise for an old disease. *Brain and Development*, **2019**, 41, 901-904	2.2	1
20	A rare variant of Guillain-Barr syndrome with acute motor axonal neuropathy (AMAN) in a Caucasian boy. *Neuropediatrics*, **2000**, 31, 162-3	1.6	1
19	Mutation screening of neurofibromatosis type 1 (NF1) exons 28 and 29 with single strand conformation polymorphism (SSCP): five novel mutations, one recurrent transition and two polymorphisms in a panel of 118 unrelated NF1 patients. Mutations in brief no. 229. Online. *Human Mutation*, **1999**, 13, 258	4.7	1
18	What can go wrong in the non-coding genome and how to interpret whole genome sequencing data. *Medizinische Genetik*, **2021**, 33, 121-131	0.5	1
17	Public data sources for regulatory genomic features. *Medizinische Genetik*, **2021**, 33, 167-177	0.5	1
16	Presence of anti-neuronal antibodies in children with neurological disorders beyond encephalitis. *European Journal of Paediatric Neurology*, **2020**, 28, 159-166	3.8	1
15	Novel bi-allelic variants expand the SPTBN4-related genetic and phenotypic spectrum. *European Journal of Human Genetics*, **2021**, 29, 1121-1128	5.3	1
14	Aicardi-Goutires syndrome with muscle involvement in early infancy. *Neuropathology and Applied Neurobiology*, **2018**, 44, 737-742	5.2	1
13	Cytoplasmic body myopathy revisited. *Neuromuscular Disorders*, **2018**, 28, 969-971	2.9	1
12	New nuclear encoded mitochondrial mutation illustrates pitfalls in prenatal diagnosis by biochemical methods. *Clinical Chemistry*, **2002**, 48, 772-5	5.5	1
11	Homozygous mutation in murine retrovirus integration site 1 gene associated with a non-syndromic form of isolated familial achalasia. *Neurogastroenterology and Motility*, **2020**, 32, e13923	4	0

List of Publications

10	Successful plasmapheresis and immunoglobulin treatment for severe lipid storage myopathy: Doing the right thing for the wrong reason. *Neuropathology and Applied Neurobiology*, **2021**,	5.2	0
9	A New Mechanism in THRA Resistance: The First Disease-Associated Variant Leading to an Increased Inhibitory Function of THRA2. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	0
8	A novel mutation in NEB causing foetal nemaline myopathy with arthrogryposis during early gestation. *Neuromuscular Disorders*, **2021**, 31, 239-245	2.9	0
7	Reply: Impaired mitochondrial function abolishes gamma oscillations in the hippocampus through an effect on fast-spiking interneurons. *Brain*, **2011**, 134, e181-e181	11.2	
6	Corrigendum to Mammalian mitochondrial nitric oxide synthase: Characterization of a novel candidate[FEBS Lett. 580 (2006) 455462]. *FEBS Letters*, **2007**, 581, 2072-2073	3.8	
5	Mutation detection in the non-coding genome. *Medizinische Genetik*, **2021**, 33, 119-120	0.5	
4	Mitochondriale Erkrankungen **2015**, A13.1-A13.5		
3	Motor Function in Pediatric ALL Survivors after Chemotherapy-Only. *Neuropediatrics*, **2017**, 48, S1-S45	1.6	
2	Generation of four iPSC lines from four patients with Leigh syndrome carrying homoplasmic mutations m.8993T>G or m.8993T>C in the mitochondrial gene MT-ATP6.. *Stem Cell Research*, **2022**, 61, 102742	1.6	
1	Diagnosis of Taenia solium infections based on "mail order" RNA-sequencing of single tapeworm egg isolates from stool samples. *PLoS Neglected Tropical Diseases*, **2021**, 15, e0009787	4.8	