Markus Schuelke

List of Publications by Year
in descending order

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177 papers	19,206 citations	28274 55 h-index	11939 134 g-index
185 all docs	185 docs citations	185 times ranked	32448 citing authors

#	Article	IF	Citations
1	Successful plasmapheresis and immunoglobulin treatment for severe lipid storage myopathy: Doing the right thing for the wrong reason. Neuropathology and Applied Neurobiology, 2022, 48, .	3.2	2
2	Synonymous mutation in adenosine triphosphatase copperâ€transporting beta causes enhanced exon skipping in Wilson disease. Hepatology Communications, 2022, 6, 1611-1619.	4.3	6
3	What is the Role of Thyroid Hormone Receptor Alpha 2 (TRÎ±2) in Human Physiology?. Experimental and Clinical Endocrinology and Diabetes, 2022, 130, 296-302.	1.2	4
4	Generation of four iPSC lines from four patients with Leigh syndrome carrying homoplasmic mutations m.8993TÂ>ÂG or m.8993TÂ>ÂC in the mitochondrial gene MT-ATP6. Stem Cell Research, 2022, 61, 102742.	0.7	4
5	Extracellular matrix remodelling is associated with muscle force increase in overloaded mouse ‹i›plantaris‹/i› muscle. Neuropathology and Applied Neurobiology, 2021, 47, 218-235.	3.2	9
6	A novel mutation in NEB causing foetal nemaline myopathy with arthrogryposis during early gestation. Neuromuscular Disorders, 2021, 31, 239-245.	0.6	4
7	Expanding the clinical and molecular spectrum of ‹scp›‹i›ATP6V1A‹/i›‹/scp› related metabolic cutis laxa. Journal of Inherited Metabolic Disease, 2021, 44, 972-986.	3.6	7
8	Novel bi-allelic variants expand the SPTBN4-related genetic and phenotypic spectrum. European Journal of Human Genetics, 2021, 29, 1121-1128.	2.8	9
9	Defective metabolic programming impairs early neuronal morphogenesis in neural cultures and an organoid model of Leigh syndrome. Nature Communications, 2021, 12, 1929.	12.8	55
10	MutationTaster2021. Nucleic Acids Research, 2021, 49, W446-W451.	14.5	122
11	Inflammation, fibrosis and skeletal muscle regeneration in LGMDR9 are orchestrated by macrophages. Neuropathology and Applied Neurobiology, 2021, 47, 856-866.	3.2	6
12	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, 5338.	4.1	8
13	What can go wrong in the non-coding genome and how to interpret whole genome sequencing data. Medizinische Genetik, 2021, 33, 121-131.	0.2	4
14	Mutation detection in the non-coding genome. Medizinische Genetik, 2021, 33, 119-120.	0.2	0
15	Public data sources for regulatory genomic features. Medizinische Genetik, 2021, 33, 167-177.	0.2	1
16	Diagnosis of Taenia solium infections based on â€œmail orderâ€•RNA-sequencing of single tapeworm egg isolates from stool samples. PLoS Neglected Tropical Diseases, 2021, 15, e0009787.	3.0	1
17	Bi-Allelic UQCRFS1 Variants Are Associated with Mitochondrial Complex III Deficiency, Cardiomyopathy, and Alopecia Totalis. American Journal of Human Genetics, 2020, 106, 102-111.	6.2	36
18	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.	1.2	10

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19	Presence of anti-neuronal antibodies in children with neurological disorders beyond encephalitis. European Journal of Paediatric Neurology, 2020, 28, 159-166.	1.6	4
20	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.	1.1	6
21	Defining the ATPome reveals cross-optimization of metabolic pathways. Nature Communications, 2020, 11, 4319.	12.8	17
22	Complement deposition at the neuromuscular junction in seronegative myasthenia gravis. Acta Neuropathologica, 2020, 139, 1119-1122.	7.7	20
23	Liveâ€imaging of revertant and therapeutically restored dystrophin in the Dmd EGFPâ€mdx mouse model for Duchenne muscular dystrophy. Neuropathology and Applied Neurobiology, 2020, 46, 602-614.	3.2	6
24	Homozygous mutation in murine retrovirus integration site 1 gene associated with a nonâ€syndromic form of isolated familial achalasia. Neurogastroenterology and Motility, 2020, 32, e13923.	3.0	2
25	Allan-Herndon-Dudley-Syndrome: Considerations about the Brain Phenotype with Implications for Treatment Strategies. Experimental and Clinical Endocrinology and Diabetes, 2020, 128, 414-422.	1.2	9
26	A spontaneous missense mutation in the chromodomain helicase DNAâ€binding protein 8 (CHD8) gene: a novel association with congenital myasthenic syndrome. Neuropathology and Applied Neurobiology, 2020, 46, 588-601.	3.2	6
27	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.	1.1	1
28	RegulationSpotter: annotation and interpretation of extratranscriptic DNA variants. Nucleic Acids Research, 2019, 47, W106-W113.	14.5	17
29	I.6Arthrogryposis multiplex congenita; new genes & old acquaintances. Neuromuscular Disorders, 2019, 29, S117.	0.6	0
30	P.286Restoration of dystrophin at critical sites of expression following exon skipping. Neuromuscular Disorders, 2019, 29, S150-S151.	0.6	0
31	Hybrid genome assembly and annotation of Danionella translucida. Scientific Data, 2019, 6, 156.	5.3	21
32	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. Brain, 2019, 142, 2948-2964.	7.6	43
33	MutationDistiller: user-driven identification of pathogenic DNA variants. Nucleic Acids Research, 2019, 47, W114-W120.	14.5	37
34	Human muscle-derived CLEC14A-positive cells regenerate muscle independent of PAX7. Nature Communications, 2019, 10, 5776.	12.8	30
35	Phenotero: Annotate as you write. Clinical Genetics, 2019, 95, 287-292.	2.0	3
36	Motor function in survivors of pediatric acute lymphoblastic leukemia treated with chemotherapy-only. European Journal of Paediatric Neurology, 2019, 23, 304-316.	1.6	8

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37	De novo mutation inELOVL1causes ichthyosis,acanthosis nigricans, hypomyelination, spastic paraplegia, high frequency deafness and optic atrophy. Journal of Medical Genetics, 2019, 56, 164-175.	3.2	54
38	Muscle Weakness, Cardiomyopathy, and L-2-Hydroxyglutaric Aciduria Associated with a Novel Recessive SLC25A4 Mutation. JIMD Reports, 2018, 43, 27-35.	1.5	7
39	Caveolin 1 Promotes Renal Water and Salt Reabsorption. Scientific Reports, 2018, 8, 545.	3.3	8
40	Morvan syndrome associated with CASPR2 and LGI1 antibodies in a child. Neurology, 2018, 90, 183-185.	1.1	15
41	Two patients with MIRAGE syndrome lacking haematological features: role of somatic second-site reversion SAMD9 mutations. Journal of Medical Genetics, 2018, 55, 81-85.	3.2	49
42	Aicardiâ€GoutiÃ¨res syndrome with muscle involvement in early infancy. Neuropathology and Applied Neurobiology, 2018, 44, 737-742.	3.2	3
43	METABOLIC MYOPATHIES I. Neuromuscular Disorders, 2018, 28, S114.	0.6	0
44	Autophagic vacuolar myopathy is a common feature of <scp>CLN</scp>3 disease. Annals of Clinical and Translational Neurology, 2018, 5, 1385-1393.	3.7	10
45	Transparent Danionella translucida as a genetically tractable vertebrate brain model. Nature Methods, 2018, 15, 977-983.	19.0	62
46	Cytoplasmic body myopathy revisited. Neuromuscular Disorders, 2018, 28, 969-971.	0.6	3
47	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. Orphanet Journal of Rare Diseases, 2018, 13, 120.	2.7	61
48	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.	6.2	65
49	Human iPSC-Derived Neural Progenitors Are an Effective Drug Discovery Model for Neurological mtDNA Disorders. Cell Stem Cell, 2017, 20, 659-674.e9.	11.1	126
50	A novelTRAPPC11mutation in two Turkish families associated with cerebral atrophy, global retardation, scoliosis, achalasia and alacrima. Journal of Medical Genetics, 2017, 54, 176-185.	3.2	44
51	Myopathology in the times of modern genetics. Neuropathology and Applied Neurobiology, 2017, 43, 44-61.	3.2	16
52	CoQ deficiency causes disruption of mitochondrial sulfide oxidation, a new pathomechanism associated with this syndrome. EMBO Molecular Medicine, 2017, 9, 78-95.	6.9	59
53	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.	6.2	56
54	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.	2.9	15

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55	International Workshop:. Neuromuscular Disorders, 2017, 27, 1126-1137.	0.6	58
56	BMP signaling regulates satellite cell dependent postnatal muscle growth. Development (Cambridge), 2017, 144, 2737-2747.	2.5	34
57	Recessive mutation in EXOSC3 associates with mitochondrial dysfunction and pontocerebellar hypoplasia. Mitochondrion, 2017, 37, 46-54.	3.4	26
58	A recessive mutation in beta-IV-spectrin (SPTBN4) associates with congenital myopathy, neuropathy, and central deafness. Human Genetics, 2017, 136, 903-910.	3.8	51
59	KlÃ¼verâ€“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.	2.8	5
60	Coenzyme Q deficiency causes impairment of the sulfide oxidation pathway. EMBO Molecular Medicine, 2017, 9, 96-111.	6.9	61
61	Motor Function in Pediatric ALL Survivors after Chemotherapy-Only. Neuropediatrics, 2017, 48, S1-S45.	0.6	0
62	Nemaline body myopathy caused by a novel mutation in troponin T1 (TNNT1). Muscle and Nerve, 2016, 53, 564-569.	2.2	39
63	BRAT1 mutations are associated with infantile epileptic encephalopathy, mitochondrial dysfunction, and survival into childhood. American Journal of Medical Genetics, Part A, 2016, 170, 2274-2281.	1.2	25
64	Altered RNA metabolism due to a homozygousRBM7mutation in a patient with spinal motor neuropathy. Human Molecular Genetics, 2016, 25, ddw149.	2.9	35
65	A systematic, large-scale comparison of transcription factor binding site models. BMC Genomics, 2016, 17, 388.	2.8	15
66	Kyphoscoliosis peptidase (KY) mutation causes a novel congenital myopathy with core targetoid defects. Acta Neuropathologica, 2016, 132, 475-478.	7.7	16
67	Characterization of a Dmd EGFP reporter mouse as a tool to investigate dystrophin expression. Skeletal Muscle, 2016, 6, 25.	4.2	17
68	A movement disorder with dystonia and ataxia caused by a mutation in the HIBCH gene. Movement Disorders, 2016, 31, 1733-1739.	3.9	35
69	MORC2 mutation causes severe spinal muscular atrophy-phenotype, cerebellar atrophy, and diaphragmatic paralysis. Brain, 2016, 139, e70-e70.	7.6	33
70	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.	6.2	56
71	Deficiency of <scp>ECHS</scp>1 causes mitochondrial encephalopathy with cardiac involvement. Annals of Clinical and Translational Neurology, 2015, 2, 492-509.	3.7	90
72	Recessive truncating IGHMBP2 mutations presenting as axonal sensorimotor neuropathy. Neurology, 2015, 84, 523-531.	1.1	22

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73	Recessive REEP1 mutation is associated with congenital axonal neuropathy and diaphragmatic palsy. Neurology: Genetics, 2015, 1, e32.	1.9	21
74	Leukodystrophy with multiple beaded periventricular cysts: unusual cranial MRI results in Canavan disease. Journal of Inherited Metabolic Disease, 2015, 38, 983-984.	3.6	6
75	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, 319-328.	6.2	83
76	RecessiveDEAF1mutation associates with autism, intellectual disability, basal ganglia dysfunction and epilepsy. Journal of Medical Genetics, 2015, 52, 607-611.	3.2	24
77	Cavin 1 function does not follow caveolar morphology. American Journal of Physiology - Cell Physiology, 2015, 308, C1023-C1030.	4.6	2
78	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.	3.4	18
79	Mitochondriale Erkrankungen. , 2015, , A13.1-A13.5.		0
80	POMKmutation in a family with congenital muscular dystrophy with merosin deficiency, hypomyelination, mild hearing deficit and intellectual disability. Journal of Medical Genetics, 2014, 51, 275-282.	3.2	52
81	Comprehensive genotyping and clinical characterisation reveal 27 novel NKX2-1 mutations and expand the phenotypic spectrum. Journal of Medical Genetics, 2014, 51, 375-387.	3.2	77
82	Potassium channel KIR4.1-specific antibodies in children with acquired demyelinating CNS disease. Neurology, 2014, 82, 470-473.	1.1	45
83	EXOSC8 mutations alter mRNA metabolism and cause hypomyelination with spinal muscular atrophy and cerebellar hypoplasia. Nature Communications, 2014, 5, 4287.	12.8	120
84	KIF1Cmutations in two families with hereditary spastic paraparesis and cerebellar dysfunction. Journal of Medical Genetics, 2014, 51, 137-142.	3.2	67
85	Identifying Dynamic Membrane Structures with Atomic-Force Microscopy and Confocal Imaging. Microscopy and Microanalysis, 2014, 20, 514-520.	0.4	7
86	MutationTaster2: mutation prediction for the deep-sequencing age. Nature Methods, 2014, 11, 361-362.	19.0	3,203
87	Dynamics of myosin degradation in intensive care unit-acquired weakness during severe critical illness. Intensive Care Medicine, 2014, 40, 528-538.	8.2	108
88	A novel frameshift mutation of C19ORF12 causes <scp>NBIA4</scp> with cerebellar atrophy and manifests with severe peripheral motor axonal neuropathy. Clinical Genetics, 2014, 85, 290-292.	2.0	9
89	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.	1.1	65
90	Infant Botulism: Is There an Association With Thiamine Deficiency?. Pediatrics, 2014, 134, e1436-e1440.	2.1	13

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91	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-R454.	1.8	65
92	Blockade of ActRIIB Signaling Triggers Muscle Fatigability and Metabolic Myopathy. Molecular Therapy, 2014, 22, 1423-1433.	8.2	63
93	G.P.195. Neuromuscular Disorders, 2014, 24, 875.	0.6	0
94	CARbon DIoxide for the treatment of Febrile seizures: rationale, feasibility, and design of the CARDIF-study. Journal of Translational Medicine, 2013, 11, 157.	4.4	16
95	Quantitative and qualitative 2D electrophoretic analysis of differentially expressed mitochondrial proteins from five mouse organs. Proteomics, 2013, 13, 179-195.	2.2	7
96	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, 681-695.	6.2	68
97	Variants in CPA1 are strongly associated with early onset chronic pancreatitis. Nature Genetics, 2013, 45, 1216-1220.	21.4	255
98	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, 529-533.	3.2	3
99	HomozygosityMapper2012–bridging the gap between homozygosity mapping and deep sequencing. Nucleic Acids Research, 2012, 40, W516-W520.	14.5	69
100	Molecular diagnosis in mitochondrial complex I deficiency using exome sequencing. Journal of Medical Genetics, 2012, 49, 277-283.	3.2	182
101	Combined Effect of AAV-U7-Induced Dystrophin Exon Skipping and Soluble Activin Type IIB Receptor in mdx Mice. Human Gene Therapy, 2012, 23, 1269-1279.	2.7	31
102	The Natural Course of Infantile Spinal Muscular Atrophy With Respiratory Distress Type 1 (SMARD1). Pediatrics, 2012, 129, e148-e156.	2.1	59
103	Mutations in MEGF10, a regulator of satellite cell myogenesis, cause early onset myopathy, areflexia, respiratory distress and dysphagia (EMARDD). Nature Genetics, 2011, 43, 1189-1192.	21.4	84
104	Synaptic PRG-1 Modulates Excitatory Transmission via Lipid Phosphate-Mediated Signaling. Cell, 2011, 146, 1043.	28.9	0
105	Region-Specific Expression of Mitochondrial Complex I Genes during Murine Brain Development. PLoS ONE, 2011, 6, e18897.	2.5	21
106	Recurrent Stroke Due to a Novel Voltage Sensor Mutation in Ca _v 2.1 Responds to Verapamil. Stroke, 2011, 42, e14-7.	2.0	39
107	NOA1 is an essential GTPase required for mitochondrial protein synthesis. Molecular Biology of the Cell, 2011, 22, 1-11.	2.1	57
108	Systematic Comparison of Three Methods for Fragmentation of Long-Range PCR Products for Next Generation Sequencing. PLoS ONE, 2011, 6, e28240.	2.5	106

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109	Reply: Impaired mitochondrial function abolishes gamma oscillations in the hippocampus through an effect on fast-spiking interneurons. Brain, 2011, 134, e181-e181.	7.6	0
110	Gamma oscillations in the hippocampus require high complex I gene expression and strong functional performance of mitochondria. Brain, 2011, 134, 345-358.	7.6	156
111	MutationTaster evaluates disease-causing potential of sequence alterations. Nature Methods, 2010, 7, 575-576.	19.0	2,538
112	Treatment of CoQ10 Deficient Fibroblasts with Ubiquinone, CoQ Analogs, and Vitamin C: Time- and Compound-Dependent Effects. PLoS ONE, 2010, 5, e11897.	2.5	92
113	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.	2.5	25
114	Reactive oxygen species, oxidative stress, and cell death correlate with level of CoQ ₁₀ deficiency. FASEB Journal, 2010, 24, 3733-3743.	0.5	142
115	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.	3.5	198
116	P3.16 Myostatin inhibits differentiation of normal and dysferlin-deficient human skeletal myoblasts â€“ similarities and differences. Neuromuscular Disorders, 2010, 20, 645.	0.6	0
117	HomozygosityMapper--an interactive approach to homozygosity mapping. Nucleic Acids Research, 2009, 37, W593-W599.	14.5	331
118	IGHMBP2 is a ribosome-associated helicase inactive in the neuromuscular disorder distal SMA type 1 (DSMA1). Human Molecular Genetics, 2009, 18, 1288-1300.	2.9	88
119	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.	3.9	43
120	Mutations in PYCR1 cause cutis laxa with progeroid features. Nature Genetics, 2009, 41, 1016-1021.	21.4	211
121	Protracted course of juvenile ceroid lipofuscinosis associated with a novel CLN3 mutation (p.Y199X). Clinical Genetics, 2009, 76, 38-45.	2.0	26
122	Synaptic PRG-1 Modulates Excitatory Transmission via Lipid Phosphate-Mediated Signaling. Cell, 2009, 138, 1222-1235.	28.9	124
123	Comparative analysis of uncoupling protein 4 distribution in various tissues under physiological conditions and during development. Biochimica Et Biophysica Acta - Biomembranes, 2009, 1788, 2309-2319.	2.6	59
124	Muscle 3243Aâ†'G mutation load and capacity of the mitochondrial energyâ€generating system. Annals of Neurology, 2008, 63, 473-481.	5.3	25
125	Familial Glucocorticoid Deficiency Type 1 due to a Novel Compound Heterozygous <i>MC2R</i> Mutation. Hormone Research in Paediatrics, 2008, 69, 363-368.	1.8	11
126	Respiratory chain dysfunction and oxidative stress correlate with severity of primary CoQ ₁₀ deficiency. FASEB Journal, 2008, 22, 1874-1885.	0.5	150

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127	Spinal Muscular Atrophy With Respiratory Distress Type 1 (SMARD1). Journal of Child Neurology, 2008, 23, 199-204.	1.4	49
128	GeneDistillerâ€"Distilling Candidate Genes from Linkage Intervals. PLoS ONE, 2008, 3, e3874.	2.5	98
129	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, 1835-1840.	7.1	341
130	Muscle and nerve pathology in Dunnigan familial partial lipodystrophy. Neurology, 2007, 68, 677-683.	1.1	29
131	Analysis of Mitochondrial DNA in Discordant Monozygotic Twins With Neurofibromatosis Type 1. Twin Research and Human Genetics, 2007, 10, 486-495.	0.6	26
132	Corrigendum to â€œMammalian mitochondrial nitric oxide synthase: Characterization of a novel candidateâ€•[FEBS Lett. 580 (2006) 455-462]. FEBS Letters, 2007, 581, 2072-2073.	2.8	0
133	Prenatal manifestation of pancytopenia in Pearson marrow-pancreas syndrome caused by a mitochondrial DNA deletion. American Journal of Medical Genetics, Part A, 2007, 143A, 285-288.	1.2	3
134	Clinical and mutational profile in spinal muscular atrophy with respiratory distress (SMARD): defining novel phenotypes through hierarchical cluster analysis. Human Mutation, 2007, 28, 808-815.	2.5	70
135	Detection of novel NF1 mutations and rapid mutation prescreening with Pyrosequencing. Electrophoresis, 2007, 28, 4295-4301.	2.4	14
136	Tandem duplication ofDMD exon 18 associated with epilepsy, macroglossia, and endocrinologic abnormalities. Muscle and Nerve, 2007, 35, 396-401.	2.2	7
137	De novo double mutation in PAX6 and mtDNA tRNA Lys associated with atypical aniridia and mitochondrial disease. Journal of Molecular Medicine, 2007, 85, 163-168.	3.9	11
138	Improved glucose metabolism in mice lacking Î±-tocopherol transfer protein. European Journal of Nutrition, 2007, 46, 397-405.	3.9	12
139	Leigh Syndrome with Nephropathy and CoQ10 Deficiency Due to decaprenyl diphosphate synthase subunit 2 (PDSS2) Mutations. American Journal of Human Genetics, 2006, 79, 1125-1129.	6.2	359
140	Mammalian mitochondrial nitric oxide synthase: Characterization of a novel candidate. FEBS Letters, 2006, 580, 455-462.	2.8	43
141	A New Mutation of IGHMBP2 Gene. Pediatric Neurology, 2006, 34, 168.	2.1	5
142	Genetic deafness in a preterm infant with a critical postnatal course. Pediatric Critical Care Medicine, 2006, 7, 270-272.	0.5	3
143	The spectrum ofWRNmutations in Werner syndrome patients. Human Mutation, 2006, 27, 558-567.	2.5	198
144	Genotypic and phenotypic spectrum of PANK2 mutations in patients with neurodegeneration with brain iron accumulation. Annals of Neurology, 2006, 59, 248-256.	5.3	184

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145	Identification of small non-coding RNAs from mitochondria and chloroplasts. Nucleic Acids Research, 2006, 34, 3842-3852.	14.5	161
146	Increased mRNA expression of tissue inhibitors of metalloproteinase-1 and -2 in Duchenne muscular dystrophy. Acta Neuropathologica, 2005, 109, 285-293.	7.7	38
147	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, 2981-2999.	2.2	23
148	Neonatal Lactic Acidosis, Complex I/IV Deficiency, and Fetal Cerebral Disruption. Neuropediatrics, 2005, 36, 193-199.	0.6	33
149	Molecular diagnostics of mitochondrial encephalomyopathies. Neuropediatrics, 2005, 36, .	0.6	0
150	Genomic rearrangements at the IGHMBP2 gene locus in two patients with SMARD1. Human Genetics, 2004, 115, 319-326.	3.8	35
151	Localization of Î±-Tocopherol Transfer Protein in Trophoblast, Fetal Capillaries' Endothelium and Amnion Epithelium of Human Term Placenta. Free Radical Research, 2004, 38, 413-420.	3.3	47
152	Myostatin Mutation Associated with Gross Muscle Hypertrophy in a Child. New England Journal of Medicine, 2004, 350, 2682-2688.	27.0	1,238
153	Mutations in the Gene Encoding Gap Junction Protein Î±12 (Connexin 46.6) Cause Pelizaeus-Merzbacherâ€“Like Disease. American Journal of Human Genetics, 2004, 75, 251-260.	6.2	257
154	Infantile spinal muscular atrophy with respiratory distress type 1 (SMARD1). Annals of Neurology, 2003, 54, 719-724.	5.3	141
155	New Nuclear Encoded Mitochondrial Mutation Illustrates Pitfalls in Prenatal Diagnosis by Biochemical Methods. Clinical Chemistry, 2002, 48, 772-775.	3.2	18
156	Septo-optic dysplasia associated with a new mitochondrialcytochrome b mutation. Annals of Neurology, 2002, 51, 388-392.	5.3	81
157	New nuclear encoded mitochondrial mutation illustrates pitfalls in prenatal diagnosis by biochemical methods. Clinical Chemistry, 2002, 48, 772-5.	3.2	1
158	Mutations in the gene encoding immunoglobulin Î¼-binding protein 2 cause spinal muscular atrophy with respiratory distress type 1. Nature Genetics, 2001, 29, 75-77.	21.4	317
159	Multiple origins of the mtDNA 7472insC mutation associated with hearing loss and neurological dysfunction. European Journal of Human Genetics, 2001, 9, 385-387.	2.8	19
160	An economic method for the fluorescent labeling of PCR fragments. Nature Biotechnology, 2000, 18, 233-234.	17.5	3,083
161	Ataxia with vitamin E deficiency: Biochemical effects of malcompliance with vitamin E therapy. Neurology, 2000, 55, 1584-1586.	1.1	14
162	A Rare Variant of Guillain-BarrÃ© Syndrome with Acute Motor Axonal Neuropathy (AMAN) in a Caucasian Boy. Neuropediatrics, 2000, 31, 162-163.	0.6	2

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163	Heterozygous myogenic factor 6 mutation associated with myopathy and severe course of Becker muscular dystrophy. Neuromuscular Disorders, 2000, 10, 572-577.	0.6	57
164	Urinary Î±-tocopherol metabolites in Î±-tocopherol transfer protein-deficient patients. Journal of Lipid Research, 2000, 41, 1543-1551.	4.2	67
165	Urinary alpha-tocopherol metabolites in alpha-tocopherol transfer protein-deficient patients. Journal of Lipid Research, 2000, 41, 1543-51.	4.2	59
166	Spinal muscular atrophy-like picture, cardiomyopathy, and cytochrome c oxidase deficiency. Neurology, 1999, 52, 383-383.	1.1	36
167	Mutant NDUFV1Âsubunit of mitochondrial complex I causes leukodystrophy and myoclonic epilepsy. Nature Genetics, 1999, 21, 260-261.	21.4	265
168	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. Human Mutation, 1999, 13, 258-258.	2.5	2
169	Treatment of ataxia in isolated vitamin E deficiency caused by Î±-tocopherol transfer protein deficiency. Journal of Pediatrics, 1999, 134, 240-244.	1.8	70
170	Epilepsia partialis continua associated with a homoplasmic mitochondrial tRNASer(UCN) mutation. Annals of Neurology, 1998, 44, 700-704.	5.3	66
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