Markus Schuelke

List of Publications by Year
in descending order

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177 papers	19,206 citations	28272 55 h-index	11937 134 g-index
185 all docs	185 docs citations	185 times ranked	32448 citing authors

#	Article	IF	Citations
1	MutationTaster2: mutation prediction for the deep-sequencing age. Nature Methods, 2014, 11, 361-362.	19.0	3,203
2	An economic method for the fluorescent labeling of PCR fragments. Nature Biotechnology, 2000, 18, 233-234.	17.5	3,083
3	MutationTaster evaluates disease-causing potential of sequence alterations. Nature Methods, 2010, 7, 575-576.	19.0	2,538
4	Myostatin Mutation Associated with Gross Muscle Hypertrophy in a Child. New England Journal of Medicine, 2004, 350, 2682-2688.	27.0	1,238
5	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
6	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
7	HomozygosityMapper--an interactive approach to homozygosity mapping. Nucleic Acids Research, 2009, 37, W593-W599.	14.5	331
8	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
9	The First Nuclear-Encoded Complex I Mutation in a Patient with Leigh Syndrome. American Journal of Human Genetics, 1998, 63, 1598-1608.	6.2	268
10	Mutant NDUFV1Âsubunit of mitochondrial complex I causes leukodystrophy and myoclonic epilepsy. Nature Genetics, 1999, 21, 260-261.	21.4	265
11	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
12	Variants in CPA1 are strongly associated with early onset chronic pancreatitis. Nature Genetics, 2013, 45, 1216-1220.	21.4	255
13	Mutations in PYCR1 cause cutis laxa with progeroid features. Nature Genetics, 2009, 41, 1016-1021.	21.4	211
14	The spectrum ofWRNmutations in Werner syndrome patients. Human Mutation, 2006, 27, 558-567.	2.5	198
15	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
16	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
17	Molecular diagnosis in mitochondrial complex I deficiency using exome sequencing. Journal of Medical Genetics, 2012, 49, 277-283.	3.2	182
18	Identification of small non-coding RNAs from mitochondria and chloroplasts. Nucleic Acids Research, 2006, 34, 3842-3852.	14.5	161

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19	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
20	Respiratory chain dysfunction and oxidative stress correlate with severity of primary CoQ ₁₀ deficiency. FASEB Journal, 2008, 22, 1874-1885.	0.5	150
21	Reactive oxygen species, oxidative stress, and cell death correlate with level of CoQ ₁₀ deficiency. FASEB Journal, 2010, 24, 3733-3743.	0.5	142
22	Infantile spinal muscular atrophy with respiratory distress type 1 (SMARD1). Annals of Neurology, 2003, 54, 719-724.	5.3	141
23	Selective disactivation of neurofibromin GAP activity in neurofibromatosis type 1. Human Molecular Genetics, 1998, 7, 1261-1268.	2.9	135
24	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
25	Synaptic PRG-1 Modulates Excitatory Transmission via Lipid Phosphate-Mediated Signaling. Cell, 2009, 138, 1222-1235.	28.9	124
26	MutationTaster2021. Nucleic Acids Research, 2021, 49, W446-W451.	14.5	122
27	EXOSC8 mutations alter mRNA metabolism and cause hypomyelination with spinal muscular atrophy and cerebellar hypoplasia. Nature Communications, 2014, 5, 4287.	12.8	120
28	Dynamics of myosin degradation in intensive care unit-acquired weakness during severe critical illness. Intensive Care Medicine, 2014, 40, 528-538.	8.2	108
29	Systematic Comparison of Three Methods for Fragmentation of Long-Range PCR Products for Next Generation Sequencing. PLoS ONE, 2011, 6, e28240.	2.5	106
30	GeneDistillerâ€"Distilling Candidate Genes from Linkage Intervals. PLoS ONE, 2008, 3, e3874.	2.5	98
31	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
32	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
33	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
34	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
35	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
36	Septo-optic dysplasia associated with a new mitochondrialcytochrome b mutation. Annals of Neurology, 2002, 51, 388-392.	5.3	81

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37	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
38	cDNA of Eight Nuclear Encoded Subunits of NADH:Ubiquinone Oxidoreductase: Human Complex I cDNA Characterization Completed. Biochemical and Biophysical Research Communications, 1998, 253, 415-422.	2.1	73
39	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
40	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
41	HomozygosityMapper2012–bridging the gap between homozygosity mapping and deep sequencing. Nucleic Acids Research, 2012, 40, W516-W520.	14.5	69
42	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
43	KIF1Cmutations in two families with hereditary spastic paraparesis and cerebellar dysfunction. Journal of Medical Genetics, 2014, 51, 137-142.	3.2	67
44	Urinary Î±-tocopherol metabolites in Î±-tocopherol transfer protein-deficient patients. Journal of Lipid Research, 2000, 41, 1543-1551.	4.2	67
45	Epilepsia partialis continua associated with a homoplasmic mitochondrial tRNASer(UCN) mutation. Annals of Neurology, 1998, 44, 700-704.	5.3	66
46	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
47	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
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	Blockade of ActRIIB Signaling Triggers Muscle Fatigability and Metabolic Myopathy. Molecular Therapy, 2014, 22, 1423-1433.	8.2	63
50	Transparent Danionella translucida as a genetically tractable vertebrate brain model. Nature Methods, 2018, 15, 977-983.	19.0	62
51	Coenzyme Q deficiency causes impairment of the sulfide oxidation pathway. EMBO Molecular Medicine, 2017, 9, 96-111.	6.9	61
52	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
53	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
54	The Natural Course of Infantile Spinal Muscular Atrophy With Respiratory Distress Type 1 (SMARD1). Pediatrics, 2012, 129, e148-e156.	2.1	59

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55	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
56	Urinary alpha-tocopherol metabolites in alpha-tocopherol transfer protein-deficient patients. Journal of Lipid Research, 2000, 41, 1543-51.	4.2	59
57	International Workshop:. Neuromuscular Disorders, 2017, 27, 1126-1137.	0.6	58
58	Heterozygous myogenic factor 6 mutation associated with myopathy and severe course of Becker muscular dystrophy. Neuromuscular Disorders, 2000, 10, 572-577.	0.6	57
59	NOA1 is an essential GTPase required for mitochondrial protein synthesis. Molecular Biology of the Cell, 2011, 22, 1-11.	2.1	57
60	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
61	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
62	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
63	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
64	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
65	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
66	Spinal Muscular Atrophy With Respiratory Distress Type 1 (SMARD1). Journal of Child Neurology, 2008, 23, 199-204.	1.4	49
67	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
68	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
69	Potassium channel KIR4.1-specific antibodies in children with acquired demyelinating CNS disease. Neurology, 2014, 82, 470-473.	1.1	45
70	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
71	Mammalian mitochondrial nitric oxide synthase: Characterization of a novel candidate. FEBS Letters, 2006, 580, 455-462.	2.8	43
72	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

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73	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. Brain, 2019, 142, 2948-2964.	7.6	43
74	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
75	Nemaline body myopathy caused by a novel mutation in troponin T1 (TNNT1). Muscle and Nerve, 2016, 53, 564-569.	2.2	39
76	Increased mRNA expression of tissue inhibitors of metalloproteinase-1 and -2 in Duchenne muscular dystrophy. Acta Neuropathologica, 2005, 109, 285-293.	7.7	38
77	MutationDistiller: user-driven identification of pathogenic DNA variants. Nucleic Acids Research, 2019, 47, W114-W120.	14.5	37
78	Spinal muscular atrophy-like picture, cardiomyopathy, and cytochrome c oxidase deficiency. Neurology, 1999, 52, 383-383.	1.1	36
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.	6.2	36
80	Liver transplantation: treatment of choice for hepatic and neurological manifestation of Wilson's disease. Clinical Transplantation, 1997, 11, 217-24.	1.6	36
81	Genomic rearrangements at the IGHMBP2 gene locus in two patients with SMARD1. Human Genetics, 2004, 115, 319-326.	3.8	35
82	Altered RNA metabolism due to a homozygousRBM7mutation in a patient with spinal motor neuropathy. Human Molecular Genetics, 2016, 25, ddw149.	2.9	35
83	A movement disorder with dystonia and ataxia caused by a mutation in the HIBCH gene. Movement Disorders, 2016, 31, 1733-1739.	3.9	35
84	BMP signaling regulates satellite cell dependent postnatal muscle growth. Development (Cambridge), 2017, 144, 2737-2747.	2.5	34
85	Neonatal Lactic Acidosis, Complex I/IV Deficiency, and Fetal Cerebral Disruption. Neuropediatrics, 2005, 36, 193-199.	0.6	33
86	MORC2 mutation causes severe spinal muscular atrophy-phenotype, cerebellar atrophy, and diaphragmatic paralysis. Brain, 2016, 139, e70-e70.	7.6	33
87	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
88	Human muscle-derived CLEC14A-positive cells regenerate muscle independent of PAX7. Nature Communications, 2019, 10, 5776.	12.8	30
89	Muscle and nerve pathology in Dunnigan familial partial lipodystrophy. Neurology, 2007, 68, 677-683.	1.1	29
90	Analysis of Mitochondrial DNA in Discordant Monozygotic Twins With Neurofibromatosis Type 1. Twin Research and Human Genetics, 2007, 10, 486-495.	0.6	26

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91	Protracted course of juvenile ceroid lipofuscinosis associated with a novel CLN3 mutation (p.Y199X). Clinical Genetics, 2009, 76, 38-45.	2.0	26
92	Recessive mutation in EXOSC3 associates with mitochondrial dysfunction and pontocerebellar hypoplasia. Mitochondrion, 2017, 37, 46-54.	3.4	26
93	Muscle 3243Aâ†'G mutation load and capacity of the mitochondrial energyâ€generating system. Annals of Neurology, 2008, 63, 473-481.	5.3	25
94	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
95	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
96	RecessiveDEAF1mutation associates with autism, intellectual disability, basal ganglia dysfunction and epilepsy. Journal of Medical Genetics, 2015, 52, 607-611.	3.2	24
97	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
98	Cloning of the Human Mitochondrial 51 kDa Subunit (NDUFV1) Reveals a 100% Antisense Homology of Its 3â€²UTR with the 5â€²UTR of the Î³-Interferon Inducible Protein (IP-30) Precursor: Is This a Link between Mitochondrial Myopathy and Inflammation?. Biochemical and Biophysical Research Communications, 1998, 245, 599-606.	2.1	22
99	Recessive truncating IGHMBP2 mutations presenting as axonal sensorimotor neuropathy. Neurology, 2015, 84, 523-531.	1.1	22
100	Region-Specific Expression of Mitochondrial Complex I Genes during Murine Brain Development. PLoS ONE, 2011, 6, e18897.	2.5	21
101	Recessive REEP1 mutation is associated with congenital axonal neuropathy and diaphragmatic palsy. Neurology: Genetics, 2015, 1, e32.	1.9	21
102	Hybrid genome assembly and annotation of Danionella translucida. Scientific Data, 2019, 6, 156.	5.3	21
103	Complement deposition at the neuromuscular junction in seronegative myasthenia gravis. Acta Neuropathologica, 2020, 139, 1119-1122.	7.7	20
104	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
105	New Nuclear Encoded Mitochondrial Mutation Illustrates Pitfalls in Prenatal Diagnosis by Biochemical Methods. Clinical Chemistry, 2002, 48, 772-775.	3.2	18
106	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
107	Characterization of a Dmd EGFP reporter mouse as a tool to investigate dystrophin expression. Skeletal Muscle, 2016, 6, 25.	4.2	17
108	RegulationSpotter: annotation and interpretation of extratranscriptic DNA variants. Nucleic Acids Research, 2019, 47, W106-W113.	14.5	17

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109	Defining the ATPome reveals cross-optimization of metabolic pathways. Nature Communications, 2020, 11, 4319.	12.8	17
110	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
111	Kyphoscoliosis peptidase (KY) mutation causes a novel congenital myopathy with core targetoid defects. Acta Neuropathologica, 2016, 132, 475-478.	7.7	16
112	Myopathology in the times of modern genetics. Neuropathology and Applied Neurobiology, 2017, 43, 44-61.	3.2	16
113	A systematic, large-scale comparison of transcription factor binding site models. BMC Genomics, 2016, 17, 388.	2.8	15
114	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
115	Morvan syndrome associated with CASPR2 and LGI1 antibodies in a child. Neurology, 2018, 90, 183-185.	1.1	15
116	New evidence for a mutation hotspot in exon 37 of theNF1 gene. , 1997, 9, 374-377.		14
117	Ataxia with vitamin E deficiency: Biochemical effects of malcompliance with vitamin E therapy. Neurology, 2000, 55, 1584-1586.	1.1	14
118	Detection of novel NF1 mutations and rapid mutation prescreening with Pyrosequencing. Electrophoresis, 2007, 28, 4295-4301.	2.4	14
119	Infant Botulism: Is There an Association With Thiamine Deficiency?. Pediatrics, 2014, 134, e1436-e1440.	2.1	13
120	Improved glucose metabolism in mice lacking Î±-tocopherol transfer protein. European Journal of Nutrition, 2007, 46, 397-405.	3.9	12
121	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
122	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
123	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
124	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
125	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
126	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

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127	Extracellular matrix remodelling is associated with muscle force increase in overloaded mouse plantaris muscle. Neuropathology and Applied Neurobiology, 2021, 47, 218-235.	3.2	9
128	Novel bi-allelic variants expand the SPTBN4-related genetic and phenotypic spectrum. European Journal of Human Genetics, 2021, 29, 1121-1128.	2.8	9
129	Caveolin 1 Promotes Renal Water and Salt Reabsorption. Scientific Reports, 2018, 8, 545.	3.3	8
130	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
131	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
132	Tandem duplication ofDMD exon 18 associated with epilepsy, macroglossia, and endocrinologic abnormalities. Muscle and Nerve, 2007, 35, 396-401.	2.2	7
133	Quantitative and qualitative 2D electrophoretic analysis of differentially expressed mitochondrial proteins from five mouse organs. Proteomics, 2013, 13, 179-195.	2.2	7
134	Identifying Dynamic Membrane Structures with Atomic-Force Microscopy and Confocal Imaging. Microscopy and Microanalysis, 2014, 20, 514-520.	0.4	7
135	Muscle Weakness, Cardiomyopathy, and L-2-Hydroxyglutaric Aciduria Associated with a Novel Recessive SLC25A4 Mutation. JIMD Reports, 2018, 43, 27-35.	1.5	7
136	Expanding the clinical and molecular spectrum of <scp>ATP6V1A</scp> related metabolic cutis laxa. Journal of Inherited Metabolic Disease, 2021, 44, 972-986.	3.6	7
137	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
138	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
139	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
140	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
141	Inflammation, fibrosis and skeletal muscle regeneration in LGMDR9 are orchestrated by macrophages. Neuropathology and Applied Neurobiology, 2021, 47, 856-866.	3.2	6
142	Synonymous mutation in adenosine triphosphatase copperâ€transporting beta causes enhanced exon skipping in Wilson disease. Hepatology Communications, 2022, 6, 1611-1619.	4.3	6
143	A New Mutation of IGHMBP2 Gene. Pediatric Neurology, 2006, 34, 168.	2.1	5
144	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

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145	Degenerative changes in unmyelinated nerve fibers in late-infantile neuronal ceroidlipofuscinosis. Acta Neuropathologica, 1998, 95, 175-183.	7.7	4
146	Presence of anti-neuronal antibodies in children with neurological disorders beyond encephalitis. European Journal of Paediatric Neurology, 2020, 28, 159-166.	1.6	4
147	A novel mutation in NEB causing foetal nemaline myopathy with arthrogryposis during early gestation. Neuromuscular Disorders, 2021, 31, 239-245.	0.6	4
148	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
149	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
150	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
151	Genetic deafness in a preterm infant with a critical postnatal course. Pediatric Critical Care Medicine, 2006, 7, 270-272.	0.5	3
152	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
153	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
154	Aicardiâ€GoutiÃ¨res syndrome with muscle involvement in early infancy. Neuropathology and Applied Neurobiology, 2018, 44, 737-742.	3.2	3
155	Cytoplasmic body myopathy revisited. Neuromuscular Disorders, 2018, 28, 969-971.	0.6	3
156	Phenotero: Annotate as you write. Clinical Genetics, 2019, 95, 287-292.	2.0	3
157	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
158	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
159	Cavin 1 function does not follow caveolar morphology. American Journal of Physiology - Cell Physiology, 2015, 308, C1023-C1030.	4.6	2
160	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
161	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
162	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

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163	Public data sources for regulatory genomic features. Medizinische Genetik, 2021, 33, 167-177.	0.2	1
164	New nuclear encoded mitochondrial mutation illustrates pitfalls in prenatal diagnosis by biochemical methods. Clinical Chemistry, 2002, 48, 772-5.	3.2	1
165	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
166	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
167	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
168	Synaptic PRG-1 Modulates Excitatory Transmission via Lipid Phosphate-Mediated Signaling. Cell, 2011, 146, 1043.	28.9	0
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