Thomas Klopstock

List of Publications by Year
in descending order

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218	16,233	15495 65	19169 118
papers	citations	h-index	g-index

232	232	232	20996
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	De Novo and Dominantly Inherited <scp>SPTAN1</scp> Mutations Cause Spastic Paraplegia and Cerebellar Ataxia. Movement Disorders, 2022, 37, 1175-1186.	2.2	9
2	DNAJC30 defect: a frequent cause of recessive Leber hereditary optic neuropathy and Leigh syndrome. Brain, 2022, 145, 1624-1631.	3.7	21
3	Lifetime risk of autosomal recessive neurodegeneration with brain iron accumulation (NBIA) disorders calculated from genetic databases. EBioMedicine, 2022, 77, 103869.	2.7	11
4	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	3.6	85
5	Community Consensus Guidelines to Support FAIR Data Standards in Clinical Research Studies in Primary Mitochondrial Disease. Genetics & Genomics Next, 2022, 3, 2100047.	0.8	1
6	Adultâ€Onset Neurodegeneration in Nucleotide Excision Repair Disorders (<scp>NERD_{ND}</scp>): Time to Move Beyond the Skin. Movement Disorders, 2022, 37, 1707-1718.	2.2	7
7	Fosmetpantotenate Randomized Controlled Trial in Pantothenate Kinaseâ€“Associated Neurodegeneration. Movement Disorders, 2021, 36, 1342-1352.	2.2	20
8	An international classification of inherited metabolic disorders (<scp>ICIMD</scp>). Journal of Inherited Metabolic Disease, 2021, 44, 164-177.	1.7	146
9	The phenotype associated with variants in <scp>TANGO2</scp> may be explained by a dual role of the protein in <scp>ERâ€toâ€Golgi</scp> transport and at the mitochondria. Journal of Inherited Metabolic Disease, 2021, 44, 426-437.	1.7	23
10	Gene Therapies for the Treatment of Leber Hereditary Optic Neuropathy. International Ophthalmology Clinics, 2021, 61, 195-208.	0.3	14
11	Treat Iron-Related Childhood-Onset Neurodegeneration (TIRCON)â€”An International Network on Care and Research for Patients With Neurodegeneration With Brain Iron Accumulation (NBIA). Frontiers in Neurology, 2021, 12, 642228.	1.1	5
12	Sulthiame impairs mitochondrial function in vitro and may trigger onset of visual loss in Leber hereditary optic neuropathy. Orphanet Journal of Rare Diseases, 2021, 16, 64.	1.2	4
13	A Nation-Wide, Multi-Center Study on the Quality of Life of ALS Patients in Germany. Brain Sciences, 2021, 11, 372.	1.1	15
14	Impaired complex I repair causes recessive Leberâ€™s hereditary optic neuropathy. Journal of Clinical Investigation, 2021, 131, .	3.9	89
15	Emerging Disease-Modifying Therapies in Neurodegeneration With Brain Iron Accumulation (NBIA) Disorders. Frontiers in Neurology, 2021, 12, 629414.	1.1	28
16	Efficacy and Safety of Intravitreal Gene Therapy for Leber Hereditary Optic Neuropathy Treated within 6 Months of Disease Onset. Ophthalmology, 2021, 128, 649-660.	2.5	87
17	Intravitreal Gene Therapy vs. Natural History in Patients With Leber Hereditary Optic Neuropathy Carrying the m.11778G>A ND4 Mutation: Systematic Review and Indirect Comparison. Frontiers in Neurology, 2021, 12, 662838.	1.1	42
18	A comprehensive phenotypic characterization of a whole-body Wdr45 knock-out mouse. Mammalian Genome, 2021, 32, 332-349.	1.0	4

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19	Rational Design of Novel Therapies for Pantothenate <scp>Kinaseâ€“Associated</scp> Neurodegeneration. Movement Disorders, 2021, 36, 2005-2016.	2.2	12
20	Progression characteristics of the European Friedreich's Ataxia Consortium for Translational Studies (EFACTS): a 4-year cohort study. Lancet Neurology, The, 2021, 20, 362-372.	4.9	53
21	The ARCA Registry: A Collaborative Global Platform for Advancing Trial Readiness in Autosomal Recessive Cerebellar Ataxias. Frontiers in Neurology, 2021, 12, 677551.	1.1	15
22	Informal Caregiving in Amyotrophic Lateral Sclerosis (ALS): A High Caregiver Burden and Drastic Consequences on Caregiversâ€™ Lives. Brain Sciences, 2021, 11, 748.	1.1	30
23	Mitochondrial disease in adults: recent advances and future promise. Lancet Neurology, The, 2021, 20, 573-584.	4.9	96
24	Cross-Sectional Analysis of Baseline Visual Parameters in Subjects Recruited Into the RESCUE and REVERSE ND4-LHON Gene Therapy Studies. Journal of Neuro-Ophthalmology, 2021, 41, 298-308.	0.4	5
25	Multi-Omics Approach to Mitochondrial DNA Damage in Human Muscle Fibers. International Journal of Molecular Sciences, 2021, 22, 11080.	1.8	2
26	Autosomal dominant optic atrophy: A novel treatment for OPA1 splice defects using U1 snRNA adaption. Molecular Therapy - Nucleic Acids, 2021, 26, 1186-1197.	2.3	8
27	Long-Term Follow-Up After Unilateral Intravitreal Gene Therapy for Leber Hereditary Optic Neuropathy: The RESTORE Study. Journal of Neuro-Ophthalmology, 2021, 41, 309-315.	0.4	30
28	Mitochondrial disorders. Deutsches Ärzteblatt International, 2021, , .	0.6	12
29	Safety and Efficacy of Acetyl-DL-Leucine in Certain Types of Cerebellar Ataxia. JAMA Network Open, 2021, 4, e2135841.	2.8	16
30	Determining histone H4 acetylation patterns in human peripheral blood mononuclear cells using mass spectrometry. Clinical Mass Spectrometry, 2020, 15, 54-60.	1.9	3
31	Brain Iron and Metabolic Abnormalities in C19orf12 Mutation Carriers: A 7.0 Tesla MRI Study in Mitochondrial Membrane Proteinâ€“Associated Neurodegeneration. Movement Disorders, 2020, 35, 142-150.	2.2	16
32	In-depth phenotyping reveals common and novel disease symptoms in a hemizygous knock-in mouse model (Mut-ko/ki) of mut-type methylmalonic aciduria. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2020, 1866, 165622.	1.8	12
33	Real-World Clinical Experience With Idebenone in the Treatment of Leber Hereditary Optic Neuropathy. Journal of Neuro-Ophthalmology, 2020, 40, 558-565.	0.4	72
34	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	4.7	43
35	Onset features and time to diagnosis in Friedreichâ€™s Ataxia. Orphanet Journal of Rare Diseases, 2020, 15, 198.	1.2	27
36	PKAN neurodegeneration and residual PANK2 activities in patient erythrocytes. Annals of Clinical and Translational Neurology, 2020, 7, 1340-1351.	1.7	11

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37	Bi-allelic HPDL Variants Cause a Neurodegenerative Disease Ranging from Neonatal Encephalopathy to Adolescent-Onset Spastic Paraplegia. American Journal of Human Genetics, 2020, 107, 364-373.	2.6	30
38	Biotinidase deficiency. Neurology: Genetics, 2020, 6, e525.	0.9	3
39	LINS1-associated neurodevelopmental disorder. Neurology: Genetics, 2020, 6, e500.	0.9	3
40	Bilateral visual improvement with unilateral gene therapy injection for Leber hereditary optic neuropathy. Science Translational Medicine, 2020, 12, .	5.8	128
41	Association of A Novel Splice Site Mutation in P/Q-Type Calcium Channels with Childhood Epilepsy and Late-Onset Slowly Progressive Non-Episodic Cerebellar Ataxia. International Journal of Molecular Sciences, 2020, 21, 3810.	1.8	14
42	Delineating MT-ATP6 -associated disease. Neurology: Genetics, 2020, 6, e393.	0.9	73
43	Physiological relevance of the neuronal isoform of inositol-1,4,5-trisphosphate 3-kinases in mice. Neuroscience Letters, 2020, 735, 135206.	1.0	3
44	A comprehensive and comparative phenotypic analysis of the collaborative founder strains identifies new and known phenotypes. Mammalian Genome, 2020, 31, 30-48.	1.0	22
45	Functional characterization of the first missense variant in CEP78 , a founder allele associated with coneâ€rod dystrophy, hearing loss, and reduced male fertility. Human Mutation, 2020, 41, 998-1011.	1.1	15
46	Clinicoâ€Genetic, Imaging and Molecular Delineation of <scp>COQ8A</scp>â€Ataxia: A Multicenter Study of 59 Patients. Annals of Neurology, 2020, 88, 251-263.	2.8	52
47	Lifetime risk of autosomal recessive mitochondrial disorders calculated from genetic databases. EBioMedicine, 2020, 54, 102730.	2.7	35
48	Clinico-Genetic, Imaging and Molecular Delineation of COQ8A-Ataxia: A Multicenter Study of 59 Patients. , 2020, 88, 251.		1
49	Diagnostic and clinical experience of patients with pantothenate kinase-associated neurodegeneration. Orphanet Journal of Rare Diseases, 2019, 14, 174.	1.2	10
50	Gillespie's Syndrome with Minor Cerebellar Involvement and No Intellectual Disability Associated with a Novel ITPR1 Mutation: Report of a Case and Literature Review. Neuropediatrics, 2019, 50, 382-386.	0.3	8
51	Erythrocyte Encapsulated Thymidine Phosphorylase for the Treatment of Patients with Mitochondrial Neurogastrointestinal Encephalomyopathy: Study Protocol for a Multi-Centre, Multiple Dose, Open Label Trial. Journal of Clinical Medicine, 2019, 8, 1096.	1.0	39
52	Protocol of a randomized, double-blind, placebo-controlled, parallel-group, multicentre study of the efficacy and safety of nicotinamide in patients with Friedreich ataxia (NICOFA). Neurological Research and Practice, 2019, 1, 33.	1.0	14
53	Reply: Adult-onset distal spinal muscular atrophy: a new phenotype associated with KIF5A mutations. Brain, 2019, 142, e67-e67.	3.7	1
54	Charles Bonnet syndrome in Leberâ€™s hereditary optic neuropathy. Journal of Neurology, 2019, 266, 777-779.	1.8	6

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55	Safety and efficacy of deferiprone for pantothenate kinase-associated neurodegeneration: a randomised, double-blind, controlled trial and an open-label extension study. Lancet Neurology, The, 2019, 18, 631-642.	4.9	102
56	Low catalytic activity is insufficient to induce disease pathology in triosephosphate isomerase deficiency. Journal of Inherited Metabolic Disease, 2019, 42, 839-849.	1.7	13
57	The FOsmetpantotenate Replacement Therapy (FORT) randomized, double-blind, Placebo-controlled pivotal trial: Study design and development methodology of a novel primary efficacy outcome in patients with pantothenate kinase-associated neurodegeneration. Clinical Trials, 2019, 16, 410-418.	0.7	13
58	Mitochondrial Symptomatic Treatments. , 2019, , 349-356.		1
59	Prognostic factors in ALS: a comparison between Germany and China. Journal of Neurology, 2019, 266, 1516-1525.	1.8	46
60	Recommendations for patient screening in ultra-rare inherited metabolic diseases: what have we learned from Niemann-Pick disease type C?. Orphanet Journal of Rare Diseases, 2019, 14, 20.	1.2	15
61	A Scale to Assess Activities of Daily Living in Pantothenate Kinaseâ€Associated Neurodegeneration. Movement Disorders Clinical Practice, 2019, 6, 139-149.	0.8	9
62	MT-ATP6mitochondrial disease variants: Phenotypic and biochemical features analysis in 218 published cases and cohort of 14 new cases. Human Mutation, 2019, 40, 499-515.	1.1	70
63	Proteomics of Cytochrome c Oxidase-Negative versus -Positive Muscle Fiber Sections in Mitochondrial Myopathy. Cell Reports, 2019, 29, 3825-3834.e4.	2.9	17
64	A mouse model for intellectual disability caused by mutations in the X-linked 2â€²â€²Oâ€‘methyltransferase Ftsj1 gene. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2019, 1865, 2083-2093.	1.8	17
65	Consensus-based statements for the management of mitochondrial stroke-like episodes. Wellcome Open Research, 2019, 4, 201.	0.9	66
66	Hot-spot KIF5A mutations cause familial ALS. Brain, 2018, 141, 688-697.	3.7	167
67	Comprehensive analysis of the mutation spectrum in 301 German ALS families. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 817-827.	0.9	80
68	Brain diffusion tensor imaging changes in cerebrotendinous xanthomatosis reversed with treatment. Journal of Neurology, 2018, 265, 388-393.	1.8	7
69	Friedreich and dominant ataxias: quantitative differences in cerebellar dysfunction measurements. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 559-565.	0.9	16
70	Fgf9 Y162C Mutation Alters Information Processing and Social Memory in Mice. Molecular Neurobiology, 2018, 55, 4580-4595.	1.9	11
71	Mitochondrial disorders of the retinal ganglion cells and the optic nerve. Mitochondrion, 2018, 42, 1-10.	1.6	18
72	The Role of Fibroblast Growth Factor-Binding Protein 1 in Skin Carcinogenesis and Inflammation. Journal of Investigative Dermatology, 2018, 138, 179-188.	0.3	23

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73	Understanding gene functions and disease mechanisms: Phenotyping pipelines in the German Mouse Clinic. Behavioural Brain Research, 2018, 352, 187-196.	1.2	31
74	Laboratory mouse housing conditions can be improved using common environmental enrichment without compromising data. PLoS Biology, 2018, 16, e2005019.	2.6	48
75	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. Orphanet Journal of Rare Diseases, 2018, 13, 120.	1.2	61
76	Atrophy in the Thalamus But Not Cerebellum Is Specific for C9orf72 FTD and ALS Patients â€“ An Atlas-Based Volumetric MRI Study. Frontiers in Aging Neuroscience, 2018, 10, 45.	1.7	40
77	Folinic acid therapy in cerebral folate deficiency: marked improvement in an adult patient. Journal of Neurology, 2017, 264, 578-582.	1.8	13
78	Effects of acetyl-DL-leucine on cerebellar ataxia (ALCAT trial): study protocol for a multicenter, multinational, randomized, double-blind, placebo-controlled, crossover phase III trial. BMC Neurology, 2017, 17, 7.	0.8	23
79	Lifetime exercise intolerance with lactic acidosis as key manifestation of novel compound heterozygous ACAD9 mutations causing complex I deficiency. Neuromuscular Disorders, 2017, 27, 473-476.	0.3	10
80	Spinal poly-GA inclusions in a C9orf72 mouse model trigger motor deficits and inflammation without neuron loss. Acta Neuropathologica, 2017, 134, 241-254.	3.9	99
81	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	3.7	85
82	Serum Response Factor (SRF) Ablation Interferes with Acute Stress-Associated Immediate and Long-Term Coping Mechanisms. Molecular Neurobiology, 2017, 54, 8242-8262.	1.9	12
83	Antibodies inhibit transmission and aggregation of C9orf72 polyâ€•<scp>GA</scp> dipeptide repeat proteins. EMBO Molecular Medicine, 2017, 9, 687-702.	3.3	70
84	Consensus clinical management guideline for pantothenate kinase-associated neurodegeneration (PKAN). Molecular Genetics and Metabolism, 2017, 120, 278-287.	0.5	64
85	International Workshop:. Neuromuscular Disorders, 2017, 27, 1126-1137.	0.3	58
86	Every-other-day feeding extends lifespan but fails to delay many symptoms of aging in mice. Nature Communications, 2017, 8, 155.	5.8	87
87	Clinical and genetic characteristics of sporadic adult-onset degenerative ataxia. Neurology, 2017, 89, 1043-1049.	1.5	45
88	Female mice lacking Pald1 exhibit endothelial cell apoptosis and emphysema. Scientific Reports, 2017, 7, 15453.	1.6	12
89	Meis1 effects on motor phenotypes and the sensorimotor system in mice. DMM Disease Models and Mechanisms, 2017, 10, 981-991.	1.2	25
90	Characterization of a Leber's hereditary optic neuropathy (LHON) family harboring two primary LHON mutations m.11778G > A and m.14484T > C of the mitochondrial DNA. Mitochondrion, 2017, 36, 15-20.	1.6	23

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91	Hereditary spastic paraplegia type 5: natural history, biomarkers and a randomized controlled trial. Brain, 2017, 140, 3112-3127.	3.7	87
92	International Consensus Statement on the Clinical and Therapeutic Management of Leber Hereditary Optic Neuropathy. Journal of Neuro-Ophthalmology, 2017, 37, 371-381.	0.4	156
93	Hereditary spastic paraplegia: Clinicogenetic lessons from 608 patients. Annals of Neurology, 2016, 79, 646-658.	2.8	218
94	Life span extension by targeting a link between metabolism and histone acetylation in Drosophila. EMBO Reports, 2016, 17, 455-469.	2.0	116
95	Mitochondrial replacement approaches: challenges for clinical implementation. Genome Medicine, 2016, 8, 126.	3.6	21
96	TDPâ€43 loss of function inhibits endosomal trafficking and alters trophic signaling in neurons. EMBO Journal, 2016, 35, 2350-2370.	3.5	76
97	Prevalence of Pompe disease in 3,076 patients with hyperCKemia and limb-girdle muscular weakness. Neurology, 2016, 87, 295-298.	1.5	60
98	SYNE1 ataxia is a common recessive ataxia with major non-cerebellar features: a large multi-centre study. Brain, 2016, 139, 1378-1393.	3.7	87
99	Viable Ednra Y129F mice feature human mandibulofacial dysostosis with alopecia (MFDA) syndrome due to the homologue mutation. Mammalian Genome, 2016, 27, 587-598.	1.0	5
100	Absence of the Autophagy Adaptor SQSTM1/p62 Causes Childhood-Onset Neurodegeneration with Ataxia, Dystonia, and Gaze Palsy. American Journal of Human Genetics, 2016, 99, 735-743.	2.6	99
101	NAXE Mutations Disrupt the Cellular NAD(P)HX Repair System and Cause a Lethal Neurometabolic Disorder of Early Childhood. American Journal of Human Genetics, 2016, 99, 894-902.	2.6	75
102	International Paediatric Mitochondrial Disease Scale. Journal of Inherited Metabolic Disease, 2016, 39, 705-712.	1.7	16
103	Progression characteristics of the European Friedreichâ€™s Ataxia Consortium for Translational Studies (EFACTS): a 2 year cohort study. Lancet Neurology, The, 2016, 15, 1346-1354.	4.9	117
104	Mitochondrial DNA Variation and Heteroplasmy in Monozygotic Twins Clinically Discordant for Multiple Sclerosis. Human Mutation, 2016, 37, 765-775.	1.1	41
105	Bi-allelic Truncating Mutations in TANGO2 Cause Infancy-Onset Recurrent Metabolic Crises with Encephalocardiomyopathy. American Journal of Human Genetics, 2016, 98, 358-362.	2.6	77
106	Reply: Parsing the differences in affected with LHON: genetic versus environmental triggers of disease conversion. Brain, 2016, 139, e18-e18.	3.7	13
107	Expanded phenotypic spectrum of the m.8344A>G â€œMERRFâ€•mutation: data from the German mitoNET registry. Journal of Neurology, 2016, 263, 961-972.	1.8	77
108	Creatine for neuroprotection in neurodegenerative disease: end of story?. Amino Acids, 2016, 48, 1929-1940.	1.2	68

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109	Generation and Standardized, Systemic Phenotypic Analysis of Pou3f3L423P Mutant Mice. PLoS ONE, 2016, 11, e0150472.	1.1	14
110	The First Scube3 Mutant Mouse Line with Pleiotropic Phenotypic Alterations. G3: Genes, Genomes, Genetics, 2016, 6, 4035-4046.	0.8	9
111	Deficiency of <scp>ECHS</scp>1 causes mitochondrial encephalopathy with cardiac involvement. Annals of Clinical and Translational Neurology, 2015, 2, 492-509.	1.7	90
112	<scp>M</scp>i<scp>R</scp>â€34a deficiency accelerates medulloblastoma formation in vivo. International Journal of Cancer, 2015, 136, 2293-2303.	2.3	40
113	MIM-Induced Membrane Bending Promotes Dendritic Spine Initiation. Developmental Cell, 2015, 33, 644-659.	3.1	84
114	Overexpression of the mitochondrial methyltransferase TFB1M in the mouse does not impact mitoribosomal methylation status or hearing. Human Molecular Genetics, 2015, 24, 7286-7294.	1.4	12
115	MTO1 mediates tissue specificity of OXPHOS defects via tRNA modification and translation optimization, which can be bypassed by dietary intervention. Human Molecular Genetics, 2015, 24, 2247-2266.	1.4	43
116	Biological and clinical characteristics of the European Friedreich's Ataxia Consortium for Translational Studies (EFACTS) cohort: a cross-sectional analysis of baseline data. Lancet Neurology, The, 2015, 14, 174-182.	4.9	159
117	Distribution of dipeptide repeat proteins in cellular models and C9orf72 mutation cases suggests link to transcriptional silencing. Acta Neuropathologica, 2015, 130, 537-555.	3.9	157
118	Analysis of mammalian gene function through broad-based phenotypic screens across a consortium of mouse clinics. Nature Genetics, 2015, 47, 969-978.	9.4	137
119	Novel ATM mutation in a German patient presenting as generalized dystonia without classical signs of ataxia-telangiectasia. Journal of Neurology, 2015, 262, 768-770.	1.8	17
120	Paranoid delusion as lead symptom in two siblings with late-onset Tayâ€“Sachs disease and a novel mutation in the HEXA gene. Journal of Neurology, 2015, 262, 1072-1073.	1.8	6
121	MRPL44 mutations cause a slowly progressive multisystem disease with childhood-onset hypertrophic cardiomyopathy. Neurogenetics, 2015, 16, 319-323.	0.7	44
122	Spinocerebellar ataxia type 36 exists in diverse populations and can be caused by a short hexanucleotide GGCCTG repeat expansion. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 986-995.	0.9	49
123	Abnormal Brain Iron Metabolism in Irp2 Deficient Mice Is Associated with Mild Neurological and Behavioral Impairments. PLoS ONE, 2014, 9, e98072.	1.1	45
124	Pleiotropic Functions for Transcription Factor Zscan10. PLoS ONE, 2014, 9, e104568.	1.1	16
125	MTO1-Deficient Mouse Model Mirrors the Human Phenotype Showing Complex I Defect and Cardiomyopathy. PLoS ONE, 2014, 9, e114918.	1.1	17
126	Mutations in GTPBP3 Cause a Mitochondrial Translation Defect Associated with Hypertrophic Cardiomyopathy, Lactic Acidosis, and Encephalopathy. American Journal of Human Genetics, 2014, 95, 708-720.	2.6	123

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127	Leberâ€™s hereditary optic neuropathy with late disease onset: clinical and molecular characteristics of 20 patients. Orphanet Journal of Rare Diseases, 2014, 9, 158.	1.2	58
128	Genetic Evidence for the Adhesion Protein IgSF9/Dasm1 to Regulate Inhibitory Synapse Development Independent of its Intracellular Domain. Journal of Neuroscience, 2014, 34, 4187-4199.	1.7	27
129	Safety and tolerability of carbamylated erythropoietin in Friedreich's ataxia. Movement Disorders, 2014, 29, 935-939.	2.2	46
130	Mitochondrial Dysfunction and Decrease in Body Weight of a Transgenic Knock-in Mouse Model for TDP-43. Journal of Biological Chemistry, 2014, 289, 10769-10784.	1.6	100
131	Targeted high-throughput sequencing identifies a TARDBP mutation as a cause of early-onset FTD without motor neuron disease. Neurobiology of Aging, 2014, 35, 1212.e1-1212.e5.	1.5	40
132	Recessive dystonia-ataxia syndrome in a Turkish family caused by a COX20 (FAM36A) mutation. Journal of Neurology, 2014, 261, 207-212.	1.8	40
133	Aberrant methylation of t<scp>RNA</scp>s links cellular stress to neuroâ€developmental disorders. EMBO Journal, 2014, 33, 2020-2039.	3.5	490
134	Novel phenotype associated with a mutation in the KCNA1(Kv1.1) gene. Frontiers in Physiology, 2014, 5, 525.	1.3	42
135	Effects of acetyl-dl-leucine in patients with cerebellar ataxia: a case series. Journal of Neurology, 2013, 260, 2556-2561.	1.8	89
136	SMC6 is an essential gene in mice, but a hypomorphic mutant in the ATPase domain has a mild phenotype with a range of subtle abnormalities. DNA Repair, 2013, 12, 356-366.	1.3	24
137	Analysis of <scp> LMNB </scp> 1 Duplications in Autosomal Dominant Leukodystrophy Provides Insights into Duplication Mechanisms and Alleleâ€Specific Expression. Human Mutation, 2013, 34, 1160-1171.	1.1	33
138	Lewy body pathology is associated with mitochondrial DNA damage in Parkinson's disease. Neurobiology of Aging, 2013, 34, 2231-2233.	1.5	46
139	New treatments for mitochondrial diseaseâ€”no time to drop our standards. Nature Reviews Neurology, 2013, 9, 474-481.	4.9	157
140	Loss-of-function mutations in MGME1 impair mtDNA replication and cause multisystemic mitochondrial disease. Nature Genetics, 2013, 45, 214-219.	9.4	198
141	Accumulation of mitochondrial DNA deletions within dopaminergic neurons triggers neuroprotective mechanisms. Brain, 2013, 136, 2369-2378.	3.7	66
142	Effects of Idebenone on Color Vision in Patients With Leber Hereditary Optic Neuropathy. Journal of Neuro-Ophthalmology, 2013, 33, 30-36.	0.4	68
143	Mitochondrial Membrane Protein-Associated Neurodegeneration (MPAN). International Review of Neurobiology, 2013, 110, 73-84.	0.9	39
144	High Mobility Group N Proteins Modulate the Fidelity of the Cellular Transcriptional Profile in a Tissue- and Variant-specific Manner. Journal of Biological Chemistry, 2013, 288, 16690-16703.	1.6	37

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145	TOM40 Mediates Mitochondrial Dysfunction Induced by Î±-Synuclein Accumulation in Parkinsonâ€™s Disease. PLoS ONE, 2013, 8, e62277.	1.1	133
146	Standardized, Systemic Phenotypic Analysis of UmodC93F and UmodA227T Mutant Mice. PLoS ONE, 2013, 8, e78337.	1.1	8
147	A Broad Phenotypic Screen Identifies Novel Phenotypes Driven by a Single Mutant Allele in Huntingtonâ€™s Disease CAG Knock-In Mice. PLoS ONE, 2013, 8, e80923.	1.1	36
148	Rapamycin extends murine lifespan but has limited effects on aging. Journal of Clinical Investigation, 2013, 123, 3272-3291.	3.9	333
149	Neurobeachin, a Regulator of Synaptic Protein Targeting, Is Associated with Body Fat Mass and Feeding Behavior in Mice and Body-Mass Index in Humans. PLoS Genetics, 2012, 8, e1002568.	1.5	33
150	What is influencing the phenotype of the common homozygous polymerase-Î³ mutation p.Ala467Thr?. Brain, 2012, 135, 3614-3626.	3.7	46
151	Creatine Protects against Excitoxicity in an In Vitro Model of Neurodegeneration. PLoS ONE, 2012, 7, e30554.	1.1	33
152	Srgap3 ^{â€“/â€“} mice present a neurodevelopmental disorder with schizophreniaâ€related intermediate phenotypes. FASEB Journal, 2012, 26, 4418-4428.	0.2	51
153	Pantothenate Kinase-Associated Neurodegeneration. Current Drug Targets, 2012, 13, 1182-1189.	1.0	13
154	Cytochromecoxidase subunit 4 isoform 2â€knockout mice show reduced enzyme activity, airway hyporeactivity, and lung pathology. FASEB Journal, 2012, 26, 3916-3930.	0.2	62
155	Targeted next generation sequencing as a diagnostic tool in epileptic disorders. Epilepsia, 2012, 53, 1387-1398.	2.6	299
156	Innovations in phenotyping of mouse models in the German Mouse Clinic. Mammalian Genome, 2012, 23, 611-622.	1.0	40
157	Molecular diagnosis in mitochondrial complex I deficiency using exome sequencing. Journal of Medical Genetics, 2012, 49, 277-283.	1.5	182
158	A high-throughput resequencing microarray for autosomal dominant spastic paraplegia genes. Neurogenetics, 2012, 13, 215-227.	0.7	5
159	Does enamelin have pleiotropic effects on organs other than the teeth? Lessons from a phenotyping screen of two enamelinâ€mutant mouse lines. European Journal of Oral Sciences, 2012, 120, 269-277.	0.7	6
160	Locomotion speed determines gait variability in cerebellar ataxia and vestibular failure. Movement Disorders, 2012, 27, 125-131.	2.2	150
161	Large-Scale Phenotyping of an Accurate Genetic Mouse Model of JNCL Identifies Novel Early Pathology Outside the Central Nervous System. PLoS ONE, 2012, 7, e38310.	1.1	56
162	A randomized placebo-controlled trial of idebenone in Leberâ€™s hereditary optic neuropathy. Brain, 2011, 134, 2677-2686.	3.7	461

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163	Mouse phenotyping. Methods, 2011, 53, 120-135.	1.9	128
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165	Expression analysis of dopaminergic neurons in Parkinsonâ€™s disease and aging links transcriptional dysregulation of energy metabolism to cell death. Acta Neuropathologica, 2011, 122, 75-86.	3.9	127
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