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	High levels of mitochondrial DNA deletions in substantia nigra neurons in aging and Parkinson disease. Nature Genetics, 2006, 38, 515-517.	9.4	1,363
2	Genome-wide, large-scale production of mutant mice by ENU mutagenesis. Nature Genetics, 2000, 25, 444-447.	9.4	658
3	A Humanized Version of Foxp2 Affects Cortico-Basal Ganglia Circuits in Mice. Cell, 2009, 137, 961-971.	13.5	555
4	Aberrant methylation of t<scp>RNA</scp>s links cellular stress to neuroâ€developmental disorders. EMBO Journal, 2014, 33, 2020-2039.	3.5	490
5	A randomized placebo-controlled trial of idebenone in Leberâ€™s hereditary optic neuropathy. Brain, 2011, 134, 2677-2686.	3.7	461
6	Phenotypic spectrum associated with mutations of the mitochondrial polymerase Â gene. Brain, 2006, 129, 1674-1684.	3.7	397
7	Post-Stroke Inhibition of Induced NADPH Oxidase Type 4 Prevents Oxidative Stress and Neurodegeneration. PLoS Biology, 2010, 8, e1000479.	2.6	377
8	Rapamycin extends murine lifespan but has limited effects on aging. Journal of Clinical Investigation, 2013, 123, 3272-3291.	3.9	333
9	Geneâ€“environment interactions in Leber hereditary optic neuropathy. Brain, 2009, 132, 2317-2326.	3.7	307
10	Targeted next generation sequencing as a diagnostic tool in epileptic disorders. Epilepsia, 2012, 53, 1387-1398.	2.6	299
11	Neuron-glia communication via EphA4/ephrin-A3 modulates LTP through glial glutamate transport. Nature Neuroscience, 2009, 12, 1285-1292.	7.1	258
12	SDHAF1, encoding a LYR complex-II specific assembly factor, is mutated in SDH-defective infantile leukoencephalopathy. Nature Genetics, 2009, 41, 654-656.	9.4	233
13	Absence of an Orphan Mitochondrial Protein, C19orf12, Causes a Distinct Clinical Subtype of Neurodegeneration with Brain Iron Accumulation. American Journal of Human Genetics, 2011, 89, 543-550.	2.6	224
14	Hereditary spastic paraplegia: Clinicogenetic lessons from 608 patients. Annals of Neurology, 2016, 79, 646-658.	2.8	218
15	Loss-of-function mutations in MGME1 impair mtDNA replication and cause multisystemic mitochondrial disease. Nature Genetics, 2013, 45, 214-219.	9.4	198
16	Genotypic and phenotypic spectrum of PANK2 mutations in patients with neurodegeneration with brain iron accumulation. Annals of Neurology, 2006, 59, 248-256.	2.8	184
17	Molecular diagnosis in mitochondrial complex I deficiency using exome sequencing. Journal of Medical Genetics, 2012, 49, 277-283.	1.5	182
18	Introducing the German Mouse Clinic: open access platform for standardized phenotyping. Nature Methods, 2005, 2, 403-404.	9.0	176

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19	Neuronal 3â€²,3,5-Triiodothyronine (T₃) Uptake and Behavioral Phenotype of Mice Deficient inMct8, the Neuronal T₃Transporter Mutated in Allanâ€“Herndonâ€“Dudley Syndrome. Journal of Neuroscience, 2009, 29, 9439-9449.	1.7	172
20	Hot-spot KIF5A mutations cause familial ALS. Brain, 2018, 141, 688-697.	3.7	167
21	Transfection of mitochondria: strategy towards a gene therapy of mitochondrial DNA diseases. Nucleic Acids Research, 1995, 23, 10-17.	6.5	165
22	REEP1 mutation spectrum and genotype/phenotype correlation in hereditary spastic paraplegia type 31. Brain, 2008, 131, 1078-1086.	3.7	163
23	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
24	New treatments for mitochondrial diseaseâ€”no time to drop our standards. Nature Reviews Neurology, 2013, 9, 474-481.	4.9	157
25	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
26	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
27	Locomotion speed determines gait variability in cerebellar ataxia and vestibular failure. Movement Disorders, 2012, 27, 125-131.	2.2	150
28	An international classification of inherited metabolic disorders (<scp>ICIMD</scp>). Journal of Inherited Metabolic Disease, 2021, 44, 164-177.	1.7	146
29	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
30	TOM40 Mediates Mitochondrial Dysfunction Induced by Î±-Synuclein Accumulation in Parkinsonâ€™s Disease. PLoS ONE, 2013, 8, e62277.	1.1	133
31	Comparison of three clinical rating scales in Friedreich ataxia (FRDA). Movement Disorders, 2009, 24, 1779-1784.	2.2	131
32	Mouse phenotyping. Methods, 2011, 53, 120-135.	1.9	128
33	Bilateral visual improvement with unilateral gene therapy injection for Leber hereditary optic neuropathy. Science Translational Medicine, 2020, 12, .	5.8	128
34	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
35	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
36	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

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37	Life span extension by targeting a link between metabolism and histone acetylation in Drosophila. EMBO Reports, 2016, 17, 455-469.	2.0	116
38	Creatine supplementation lowers brain glutamate levels in Huntington?s disease. Journal of Neurology, 2005, 252, 36-41.	1.8	112
39	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
40	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
41	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
42	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
43	Mitochondrial disease in adults: recent advances and future promise. Lancet Neurology, The, 2021, 20, 573-584.	4.9	96
44	Requirement of the RNA-editing Enzyme ADAR2 for Normal Physiology in Mice. Journal of Biological Chemistry, 2011, 286, 18614-18622.	1.6	91
45	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
46	Effects of acetyl-dl-leucine in patients with cerebellar ataxia: a case series. Journal of Neurology, 2013, 260, 2556-2561.	1.8	89
47	Impaired complex I repair causes recessive Leberâ€™s hereditary optic neuropathy. Journal of Clinical Investigation, 2021, 131, .	3.9	89
48	Quality of Life in Patients with Leber Hereditary Optic Neuropathy. , 2009, 50, 3112.		87
49	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
50	Every-other-day feeding extends lifespan but fails to delay many symptoms of aging in mice. Nature Communications, 2017, 8, 155.	5.8	87
51	Hereditary spastic paraplegia type 5: natural history, biomarkers and a randomized controlled trial. Brain, 2017, 140, 3112-3127.	3.7	87
52	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
53	Creatine improves health and survival of mice. Neurobiology of Aging, 2008, 29, 1404-1411.	1.5	85
54	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	3.7	85

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55	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	3.6	85
56	MIM-Induced Membrane Bending Promotes Dendritic Spine Initiation. Developmental Cell, 2015, 33, 644-659.	3.1	84
57	Comprehensive analysis of the mutation spectrum in 301 German ALS families. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 817-827.	0.9	80
58	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
59	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
60	TDPâ€43 loss of function inhibits endosomal trafficking and alters trophic signaling in neurons. EMBO Journal, 2016, 35, 2350-2370.	3.5	76
61	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
62	Mapping Gene Associations in Human Mitochondria using Clinical Disease Phenotypes. PLoS Computational Biology, 2009, 5, e1000374.	1.5	74
63	Selfâ€rated health status in spinocerebellar ataxiaâ€”Results from a European multicenter study. Movement Disorders, 2010, 25, 587-595.	2.2	74
64	Delineating MT-ATP6 -associated disease. Neurology: Genetics, 2020, 6, e393.	0.9	73
65	Dopaminergic midbrain neurons are the prime target for mitochondrial DNA deletions. Journal of Neurology, 2008, 255, 1231-1235.	1.8	72
66	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
67	Antibodies inhibit transmission and aggregation of C9orf72 polyâ€•<scp>GA</scp> dipeptide repeat proteins. EMBO Molecular Medicine, 2017, 9, 687-702.	3.3	70
68	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
69	Systemic First-Line Phenotyping. Methods in Molecular Biology, 2009, 530, 463-509.	0.4	70
70	MitoP2: An Integrative Tool for the Analysis of the Mitochondrial Proteome. Molecular Biotechnology, 2008, 40, 306-315.	1.3	69
71	Depression comorbidity in spinocerebellar ataxia. Movement Disorders, 2011, 26, 870-876.	2.2	69
72	Effects of Idebenone on Color Vision in Patients With Leber Hereditary Optic Neuropathy. Journal of Neuro-Ophthalmology, 2013, 33, 30-36.	0.4	68

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73	Creatine for neuroprotection in neurodegenerative disease: end of story?. Amino Acids, 2016, 48, 1929-1940.	1.2	68
74	Accumulation of mitochondrial DNA deletions within dopaminergic neurons triggers neuroprotective mechanisms. Brain, 2013, 136, 2369-2378.	3.7	66
75	Consensus-based statements for the management of mitochondrial stroke-like episodes. Wellcome Open Research, 2019, 4, 201.	0.9	66
76	Consensus clinical management guideline for pantothenate kinase-associated neurodegeneration (PKAN). Molecular Genetics and Metabolism, 2017, 120, 278-287.	0.5	64
77	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
78	Long-term creatine supplementation is safe in aged patients with Parkinson disease. Nutrition Research, 2008, 28, 172-178.	1.3	61
79	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
80	Drug-induced myopathies. Current Opinion in Neurology, 2008, 21, 590-595.	1.8	60
81	Prevalence of Pompe disease in 3,076 patients with hyperCKemia and limb-girdle muscular weakness. Neurology, 2016, 87, 295-298.	1.5	60
82	Iron homeostasis in the brain: complete iron regulatory protein 2 deficiency without symptomatic neurodegeneration in the mouse. Nature Genetics, 2006, 38, 967-969.	9.4	58
83	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
84	International Workshop:. Neuromuscular Disorders, 2017, 27, 1126-1137.	0.3	58
85	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
86	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
87	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
88	Srgap3 ^{â€“/â€“} mice present a neurodevelopmental disorder with schizophreniaâ€related intermediate phenotypes. FASEB Journal, 2012, 26, 4418-4428.	0.2	51
89	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
90	Laboratory mouse housing conditions can be improved using common environmental enrichment without compromising data. PLoS Biology, 2018, 16, e2005019.	2.6	48

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91	What is influencing the phenotype of the common homozygous polymerase-Î³ mutation p.Ala467Thr?. Brain, 2012, 135, 3614-3626.	3.7	46
92	Lewy body pathology is associated with mitochondrial DNA damage in Parkinson's disease. Neurobiology of Aging, 2013, 34, 2231-2233.	1.5	46
93	Safety and tolerability of carbamylated erythropoietin in Friedreich's ataxia. Movement Disorders, 2014, 29, 935-939.	2.2	46
94	Prognostic factors in ALS: a comparison between Germany and China. Journal of Neurology, 2019, 266, 1516-1525.	1.8	46
95	Abnormal Brain Iron Metabolism in Irp2 Deficient Mice Is Associated with Mild Neurological and Behavioral Impairments. PLoS ONE, 2014, 9, e98072.	1.1	45
96	Clinical and genetic characteristics of sporadic adult-onset degenerative ataxia. Neurology, 2017, 89, 1043-1049.	1.5	45
97	MRPL44 mutations cause a slowly progressive multisystem disease with childhood-onset hypertrophic cardiomyopathy. Neurogenetics, 2015, 16, 319-323.	0.7	44
98	Genomic duplications mediate overexpression of lamin B1 in adult-onset autosomal dominant leukodystrophy (ADLD) with autonomic symptoms. Neurogenetics, 2011, 12, 65-72.	0.7	43
99	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
100	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
101	Novel phenotype associated with a mutation in the KCNA1(Kv1.1) gene. Frontiers in Physiology, 2014, 5, 525.	1.3	42
102	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
103	Mitochondrial DNA Variation and Heteroplasmy in Monozygotic Twins Clinically Discordant for Multiple Sclerosis. Human Mutation, 2016, 37, 765-775.	1.1	41
104	Tongue force analysis assesses motor phenotype in premanifest and symptomatic Huntington's disease. Movement Disorders, 2010, 25, 2195-2202.	2.2	40
105	Grasping premanifest Huntington's disease â€“ shaping new endpoints for new trials. Movement Disorders, 2010, 25, 2858-2862.	2.2	40
106	Innovations in phenotyping of mouse models in the German Mouse Clinic. Mammalian Genome, 2012, 23, 611-622.	1.0	40
107	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
108	Recessive dystonia-ataxia syndrome in a Turkish family caused by a COX20 (FAM36A) mutation. Journal of Neurology, 2014, 261, 207-212.	1.8	40

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109	<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
110	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
111	Late-onset ptosis and myopathy in a patient with a heterozygous insertion in POLG2. Journal of Neurology, 2010, 257, 1517-1523.	1.8	39
112	Mitochondrial Membrane Protein-Associated Neurodegeneration (MPAN). International Review of Neurobiology, 2013, 110, 73-84.	0.9	39
113	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
114	Identification of rare DNA variants in mitochondrial disorders with improved array-based sequencing. Nucleic Acids Research, 2011, 39, 44-58.	6.5	37
115	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
116	Microphthalmia, parkinsonism, and enhanced nociception in Pitx3 416insG mice. Mammalian Genome, 2010, 21, 13-27.	1.0	36
117	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
118	Heteroplasmic mutation in the anticodon-stem of mitochondrial tRNAVal causing MNGIE-like gastrointestinal dysmotility and cachexia. Journal of Neurology, 2009, 256, 810-815.	1.8	35
119	Lifetime risk of autosomal recessive mitochondrial disorders calculated from genetic databases. EBioMedicine, 2020, 54, 102730.	2.7	35
120	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
121	Creatine Protects against Excitoxicity in an In Vitro Model of Neurodegeneration. PLoS ONE, 2012, 7, e30554.	1.1	33
122	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
123	Systematic, standardized and comprehensive neurological phenotyping of inbred mice strains in the German Mouse Clinic. Journal of Neuroscience Methods, 2006, 157, 82-90.	1.3	32
124	Understanding gene functions and disease mechanisms: Phenotyping pipelines in the German Mouse Clinic. Behavioural Brain Research, 2018, 352, 187-196.	1.2	31
125	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
126	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

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127	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
128	Clinical and molecular characterisation of a Parkinson family with a novel PINK1 mutation. Journal of Neurology, 2008, 255, 643-648.	1.8	29
129	Localization, analysis and evolution of transposed human immunoglobulin VÎº genes. Gene, 1988, 69, 215-223.	1.0	28
130	Emerging Disease-Modifying Therapies in Neurodegeneration With Brain Iron Accumulation (NBIA) Disorders. Frontiers in Neurology, 2021, 12, 629414.	1.1	28
131	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
132	Onset features and time to diagnosis in Friedreichâ€™s Ataxia. Orphanet Journal of Rare Diseases, 2020, 15, 198.	1.2	27
133	Meis1 effects on motor phenotypes and the sensorimotor system in mice. DMM Disease Models and Mechanisms, 2017, 10, 981-991.	1.2	25
134	The mouse Trm1-like gene is expressed in neural tissues and plays a role in motor coordination and exploratory behaviour. Gene, 2007, 389, 174-185.	1.0	24
135	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
136	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
137	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
138	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
139	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
140	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
141	Mitochondrial replacement approaches: challenges for clinical implementation. Genome Medicine, 2016, 8, 126.	3.6	21
142	DNAJC30 defect: a frequent cause of recessive Leber hereditary optic neuropathy and Leigh syndrome. Brain, 2022, 145, 1624-1631.	3.7	21
143	The role of complex I genes in MELAS: A novel heteroplasmic mutation 3380G>A in ND1 of mtDNA. Neuromuscular Disorders, 2008, 18, 553-556.	0.3	20
144	Fosmetpantotenate Randomized Controlled Trial in Pantothenate Kinaseâ€“Associated Neurodegeneration. Movement Disorders, 2021, 36, 1342-1352.	2.2	20

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145	Diffusion-Weighted Magnetic Resonance Imaging During the Aura of Pseudomigraine With Temporary Neurologic Symptoms and Lymphocytic Pleocytosis. Headache, 2002, 42, 294-296.	1.8	19
146	Chronic Progressive External Ophthalmoplegia: MR Spectroscopy and MR Diffusion Studies in the Brain. American Journal of Roentgenology, 2006, 187, 820-824.	1.0	19
147	Mitochondrial disorders of the retinal ganglion cells and the optic nerve. Mitochondrion, 2018, 42, 1-10.	1.6	18
148	Screening of hereditary spastic paraplegia patients for alterations at NIPA1 mutational hotspots. Journal of the Neurological Sciences, 2008, 268, 131-135.	0.3	17
149	Dll1 Haploinsufficiency in Adult Mice Leads to a Complex Phenotype Affecting Metabolic and Immunological Processes. PLoS ONE, 2009, 4, e6054.	1.1	17
150	MTO1-Deficient Mouse Model Mirrors the Human Phenotype Showing Complex I Defect and Cardiomyopathy. PLoS ONE, 2014, 9, e114918.	1.1	17
151	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
152	Proteomics of Cytochrome c Oxidase-Negative versus -Positive Muscle Fiber Sections in Mitochondrial Myopathy. Cell Reports, 2019, 29, 3825-3834.e4.	2.9	17
153	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
154	Generation of Nâ€ethylâ€Nâ€nitrosoureaâ€induced mouse mutants with deviations in plasma enzyme activities as novel organâ€specific disease models. Experimental Physiology, 2009, 94, 412-421.	0.9	16
155	Pleiotropic Functions for Transcription Factor Zscan10. PLoS ONE, 2014, 9, e104568.	1.1	16
156	International Paediatric Mitochondrial Disease Scale. Journal of Inherited Metabolic Disease, 2016, 39, 705-712.	1.7	16
157	Friedreich and dominant ataxias: quantitative differences in cerebellar dysfunction measurements. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 559-565.	0.9	16
158	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
159	Safety and Efficacy of Acetyl-DL-Leucine in Certain Types of Cerebellar Ataxia. JAMA Network Open, 2021, 4, e2135841.	2.8	16
160	Mitochondrial disorders. Current Opinion in Neurology, 2001, 14, 553-560.	1.8	15
161	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
162	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

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163	A Nation-Wide, Multi-Center Study on the Quality of Life of ALS Patients in Germany. Brain Sciences, 2021, 11, 372.	1.1	15
164	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
165	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
166	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
167	Gene Therapies for the Treatment of Leber Hereditary Optic Neuropathy. International Ophthalmology Clinics, 2021, 61, 195-208.	0.3	14
168	Generation and Standardized, Systemic Phenotypic Analysis of Pou3f3L423P Mutant Mice. PLoS ONE, 2016, 11, e0150472.	1.1	14
169	Pantothenate Kinase-Associated Neurodegeneration. Current Drug Targets, 2012, 13, 1182-1189.	1.0	13
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