H A Jinnah

List of Publications by Year
in descending order

194 papers	10,510 citations	31976 53 h-index	42399 92 g-index
198 all docs	198 docs citations	198 times ranked	7091 citing authors

#	Article	IF	Citations
1	Intranuclear inclusions and neuritic aggregates in transgenic mice expressing a mutant N-terminal fragment of huntingtin [published erratum appears in Hum Mol Genet 1999 May;8(5):943]. Human Molecular Genetics, 1999, 8, 397-407.	2.9	687
2	The functional neuroanatomy of dystonia. Neurobiology of Disease, 2011, 42, 185-201.	4.4	397
3	Definition and classification of hyperkinetic movements in childhood. Movement Disorders, 2010, 25, 1538-1549.	3.9	374
4	Grafting fibroblasts genetically modified to produce L-dopa in a rat model of Parkinson disease.. Proceedings of the National Academy of Sciences of the United States of America, 1989, 86, 9011-9014.	7.1	322
5	Survival and function of intrastriatally grafted primary fibroblasts genetically modified to produce l-dopa. Neuron, 1991, 6, 371-380.	8.1	280
6	The basal ganglia and cerebellum interact in the expression of dystonic movement. Brain, 2008, 131, 2499-2509.	7.6	280
7	Delineation of the motor disorder of Leschâ€“Nyhan disease. Brain, 2006, 129, 1201-1217.	7.6	247
8	The spectrum of inherited mutations causing HPRT deficiency: 75 new cases and a review of 196 previously reported cases. Mutation Research - Reviews in Mutation Research, 2000, 463, 309-326.	5.5	221
9	Dystonia as a network disorder: What is the role of the cerebellum?. Neuroscience, 2014, 260, 23-35.	2.3	215
10	Abnormal Cerebellar Signaling Induces Dystonia in Mice. Journal of Neuroscience, 2002, 22, 7825-7833.	3.6	189
11	The focal dystonias: Current views and challenges for future research. Movement Disorders, 2013, 28, 926-943.	3.9	184
12	Current Opinions and Areas of Consensus on the Role of the Cerebellum in Dystonia. Cerebellum, 2017, 16, 577-594.	2.5	184
13	Leschâ€“Nyhan disease and the basal ganglia. Brain Research Reviews, 2000, 32, 449-475.	9.0	166
14	Behavioral aspects of Leschâ€“Nyhan disease and its variants. Developmental Medicine and Child Neurology, 2005, 47, 673.	2.1	159
15	Dystonia rating scales: Critique and recommendations. Movement Disorders, 2013, 28, 874-883.	3.9	150
16	Research priorities in spasmodic dysphonia. Otolaryngology - Head and Neck Surgery, 2008, 139, 495-505.	1.9	147
17	Attenuated variants of Lesch-Nyhan disease. Brain, 2010, 133, 671-689.	7.6	147
18	The Anatomical Basis for Dystonia: The Motor Network Model. Tremor and Other Hyperkinetic Movements, 2020, 7, 506.	2.0	135

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19	Dopamine deficiency in a genetic mouse model of Lesch-Nyhan disease. Journal of Neuroscience, 1994, 14, 1164-1175.	3.6	131
20	A new twist on the anatomy of dystonia: The basal ganglia and the cerebellum?. Neurology, 2006, 67, 1740-1741.	1.1	129
21	Serum uric acid and brain ischemia in normal elderly adults. Neurology, 2007, 69, 1418-1423.	1.1	121
22	Diagnosis and Treatment of Dystonia. Neurologic Clinics, 2015, 33, 77-100.	1.8	120
23	Selfâ€injurious behavior: Geneâ€“brainâ€“behavior relationships. Mental Retardation and Developmental Disabilities Research Reviews, 2001, 7, 3-12.	3.6	119
24	Neuropathology of cervical dystonia. Experimental Neurology, 2013, 241, 95-104.	4.1	110
25	Genotypeâ€“phenotype correlations in neurogenetics: Lesch-Nyhan disease as a model disorder. Brain, 2014, 137, 1282-1303.	7.6	105
26	Serum uric acid and cognitive function in community-dwelling older adults.. Neuropsychology, 2007, 21, 136-140.	1.3	100
27	Functional analysis of dopaminergic systems in a DYT1 knock-in mouse model of dystonia. Neurobiology of Disease, 2012, 48, 66-78.	4.4	95
28	Shortage of Cellular ATP as a Cause of Diseases and Strategies to Enhance ATP. Frontiers in Pharmacology, 2019, 10, 98.	3.5	91
29	Assessment of patients with isolated or combined dystonia: An update on dystonia syndromes. Movement Disorders, 2013, 28, 889-898.	3.9	88
30	Treatment strategies for dystonia. Expert Opinion on Pharmacotherapy, 2010, 11, 5-15.	1.8	83
31	Development and validation of a clinical guideline for diagnosing blepharospasm. Neurology, 2013, 81, 236-240.	1.1	81
32	The Anatomical Basis for Dystonia: The Motor Network Model. Tremor and Other Hyperkinetic Movements, 2017, 7, 506.	2.0	80
33	Animal models of generalized dystonia. NeuroRx, 2005, 2, 504-512.	6.0	79
34	Treatable inherited rare movement disorders. Movement Disorders, 2018, 33, 21-35.	3.9	79
35	Hypoxanthine-guanine phosphoribosyl transferase regulates early developmental programming of dopamine neurons: implications for Lesch-Nyhan disease pathogenesis. Human Molecular Genetics, 2009, 18, 2317-2327.	2.9	78
36	Rodent models for dystonia research: Characteristics, evaluation, and utility. Movement Disorders, 2005, 20, 283-292.	3.9	77

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37	Psychiatric associations of adult-onset focal dystonia phenotypes. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 595-602.	1.9	76
38	Craniocerebral Magnetic Resonance Imaging Measurement and Findings in Lesch-Nyhan Syndrome. Archives of Neurology, 1998, 55, 547.	4.5	75
39	Limited regional cerebellar dysfunction induces focal dystonia in mice. Neurobiology of Disease, 2013, 49, 200-210.	4.4	73
40	Calcium channel agonists and dystonia in the mouse. Movement Disorders, 2000, 15, 542-551.	3.9	71
41	Cerebral Ischemia Mediates the Effect of Serum Uric Acid on Cognitive Function. Stroke, 2008, 39, 3418-3420.	2.0	71
42	Deep brain stimulation for dystonia: a novel perspective on the value of genetic testing. Journal of Neural Transmission, 2017, 124, 417-430.	2.8	68
43	The partnership of patient advocacy groups and clinical investigators in the rare diseases clinical research network. Orphanet Journal of Rare Diseases, 2016, 11, 66.	2.7	62
44	Triggers of paroxysmal dyskinesia in the calcium channel mouse mutant tottering. Pharmacology Biochemistry and Behavior, 2002, 73, 631-637.	2.9	61
45	Update on the Phenotypic Spectrum of Lesch-Nyhan Disease and its Attenuated Variants. Current Rheumatology Reports, 2012, 14, 189-194.	4.7	61
46	Metabolic disorders of purine metabolism affecting the nervous system. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1827-1836.	1.8	61
47	Abnormal motor behavior and vestibular dysfunction in the stargazer mouse mutant. Neuroscience, 2004, 127, 785-796.	2.3	60
48	Lesch-Nyhan disease: from mechanism to model and back again. DMM Disease Models and Mechanisms, 2009, 2, 116-121.	2.4	60
49	Secondary blepharospasm associated with structural lesions of the brain. Journal of the Neurological Sciences, 2013, 331, 98-101.	0.6	59
50	Irregularity distinguishes limb tremor in cervical dystonia from essential tremor. Journal of Neurology, Neurosurgery and Psychiatry, 2008, 79, 187-189.	1.9	58
51	Calcium channel activation and self-biting in mice. Proceedings of the National Academy of Sciences of the United States of America, 1999, 96, 15228-15232.	7.1	57
52	The Spectrum of Mutations Causing HPRT Deficiency: An Update. Nucleosides, Nucleotides and Nucleic Acids, 2004, 23, 1153-1160.	1.1	56
53	Neurocognitive functioning in Lesch-Nyhan disease and partial hypoxanthine-guanine phosphoribosyltransferase deficiency. Journal of the International Neuropsychological Society, 2001, 7, 805-812.	1.8	55
54	Functional analysis of brain dopamine systems in a genetic mouse model of Lesch-Nyhan syndrome. Journal of Pharmacology and Experimental Therapeutics, 1992, 263, 596-607.	2.5	55

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55	Influence of Age and Strain on Striatal Dopamine Loss in a Genetic Mouse Model of Leschâ€Nyhan Disease. Journal of Neurochemistry, 1999, 72, 225-229.	3.9	54
56	Subtle microstructural changes of the striatum in a DYT1 knock-in mouse model of dystonia. Neurobiology of Disease, 2013, 54, 362-371.	4.4	53
57	Keeping Your Head On Target. Journal of Neuroscience, 2013, 33, 11281-11295.	3.6	53
58	Botulinum toxin treatment failures in cervical dystonia: causes, management, and outcomes. Journal of Neurology, 2016, 263, 1188-1194.	3.6	53
59	Brain Purines in a Genetic Mouse Model of Lesch-Nyhan Disease. Journal of Neurochemistry, 1993, 60, 2036-2045.	3.9	51
60	Dystonia treatment. Neurology, 2017, 88, 543-550.	1.1	50
61	Leschâ€“Nyhan disease in a female with a clinically normal monozygotic twin. Molecular Genetics and Metabolism, 2005, 85, 70-77.	1.1	49
62	Clinical severity in Leschâ€“Nyhan disease: The role of residual enzyme and compensatory pathways. Molecular Genetics and Metabolism, 2015, 114, 55-61.	1.1	49
63	Dystonia genes and their biological pathways. Neurobiology of Disease, 2019, 129, 159-168.	4.4	49
64	Basal ganglia dopamine loss due to defect in purine recycling. Neurobiology of Disease, 2007, 26, 396-407.	4.4	48
65	Oscillatory head movements in cervical dystonia: Dystonia, tremor, or both?. Movement Disorders, 2015, 30, 834-842.	3.9	48
66	Distinct Behavioral and Neuropathological Abnormalities in Transgenic Mouse Models of HD and DRPLA. Neurobiology of Disease, 2001, 8, 405-418.	4.4	47
67	Consensus-Based Attributes for Identifying Patients With Spasmodic Dysphonia and Other Voice Disorders. JAMA Otolaryngology - Head and Neck Surgery, 2018, 144, 657.	2.2	47
68	Paroxysmal dyskinesias in mice. Movement Disorders, 2008, 23, 259-264.	3.9	46
69	Subtle microstructural changes of the cerebellum in a knock-in mouse model of DYT1 dystonia. Neurobiology of Disease, 2014, 62, 372-380.	4.4	46
70	Longitudinal studies of botulinum toxin in cervical dystonia: Why do patients discontinue therapy?. Toxicon, 2018, 147, 89-95.	1.6	46
71	Animal models of Lesch-Nyhan syndrome. Brain Research Bulletin, 1990, 25, 467-475.	3.0	45
72	Amphetamine-induced behavioral phenotype in a hypoxanthine-guanine phosphoribosyltransferase-deficient mouse model of Lesch-Nyhan syndrome.. Behavioral Neuroscience, 1991, 105, 1004-1012.	1.2	45

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73	Alterations of restingâ€state fMRI measurements in individuals with cervical dystonia. Human Brain Mapping, 2017, 38, 4098-4108.	3.6	45
74	Oxidative stress and dopamine deficiency in a genetic mouse model of Leschâ€“Nyhan disease. Developmental Brain Research, 2002, 133, 127-139.	1.7	44
75	Clinical and genetic features of cervical dystonia in a large multicenter cohort. Neurology: Genetics, 2016, 2, e69.	1.9	44
76	Chronic meningitis with cranial neuropathies in Wegener's granulomatosis. Case report and review of the literature. Arthritis and Rheumatism, 1997, 40, 573-577.	6.7	43
77	Evolving concepts in the pathogenesis of dystonia. Parkinsonism and Related Disorders, 2018, 46, S62-S65.	2.2	42
78	Genotype-Phenotype Correlations in Lesch-Nyhan Disease. Journal of Biological Chemistry, 2012, 287, 2997-3008.	3.4	41
79	The New Classification System for the Dystonias: Why Was It Needed and How Was It Developed?. Movement Disorders Clinical Practice, 2014, 1, 280-284.	1.5	41
80	Loss of dopamine phenotype among midbrain neurons in <scp>L</scp>eschâ€“<scp>N</scp>yhan disease. Annals of Neurology, 2014, 76, 95-107.	5.3	41
81	Clinical and demographic characteristics related to onset site and spread of cervical dystonia. Movement Disorders, 2016, 31, 1874-1882.	3.9	39
82	Regional brain volume abnormalities in Lesch-Nyhan disease and its variants: a cross-sectional study. Lancet Neurology, The, 2013, 12, 1151-1158.	10.2	38
83	Purine metabolism during neuronal differentiation: the relevance of purine synthesis and recycling. Journal of Neurochemistry, 2013, 127, 805-818.	3.9	38
84	Chapter 1 Grafting genetically modified cells to the brain: conceptual and technical issues. Progress in Brain Research, 1990, 82, 1-10.	1.4	37
85	Selective and Sustained Î±-Amino-3-hydroxy-5-methyl-4-isoxazolepropionic Acid Receptor Activation in Cerebellum Induces Dystonia in Mice. Journal of Pharmacology and Experimental Therapeutics, 2012, 340, 733-741.	2.5	37
86	Consequences of impaired purine recycling in dopaminergic neurons. Neuroscience, 2008, 152, 761-772.	2.3	36
87	How long does it take to diagnose cervical dystonia?. Journal of the Neurological Sciences, 2013, 335, 72-74.	0.6	35
88	Genotypic and phenotypic spectrum in attenuated variants of Leschâ€“Nyhan disease. Molecular Genetics and Metabolism, 2014, 112, 280-285.	1.1	35
89	Cp/Heph mutant mice have ironâ€induced neurodegeneration diminished by deferiprone. Journal of Neurochemistry, 2015, 135, 958-974.	3.9	35
90	Research Priorities in Limb and Task-Specific Dystonias. Frontiers in Neurology, 2017, 8, 170.	2.4	34

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91	HPRT Deficiency Coordinately Dysregulates Canonical Wnt and Presenilin-1 Signaling: A Neuro-Developmental Regulatory Role for a Housekeeping Gene?. PLoS ONE, 2011, 6, e16572.	2.5	34
92	Ocular motor dysfunction in lesch-Nyhan disease. Pediatric Neurology, 2001, 24, 200-204.	2.1	33
93	Paroxysmal Dyskinesias in the Lethargic Mouse Mutant. Journal of Neuroscience, 2002, 22, 8193-8200.	3.6	33
94	Experimental Therapeutics for Dystonia. Neurotherapeutics, 2008, 5, 198-209.	4.4	33
95	A Golgi study of neuronal architecture in a genetic mouse model for Leschâ€“Nyhan disease. Neurobiology of Disease, 2005, 20, 479-490.	4.4	31
96	A human neuronal tissue culture model for Lesch-Nyhan disease. Journal of Neurochemistry, 2007, 101, 841-853.	3.9	30
97	Treatment of myoclonus-dystonia syndrome with tetrabenazine. Parkinsonism and Related Disorders, 2014, 20, 1423-1426.	2.2	30
98	Brain white matter volume abnormalities in Lesch-Nyhan disease and its variants. Neurology, 2015, 84, 190-196.	1.1	30
99	Severe gouty arthritis and mild neurologic symptoms due to F199C, a newly identified variant of the hypoxanthine guanine phosphoribosyltransferase. Arthritis and Rheumatism, 2009, 60, 2201-2204.	6.7	29
100	A Functional Magnetic Resonance Imaging Study of Head Movements in Cervical Dystonia. Frontiers in Neurology, 2016, 7, 201.	2.4	29
101	The role of pallidum in the neural integrator model of cervical dystonia. Neurobiology of Disease, 2019, 125, 45-54.	4.4	29
102	Trihexyphenidyl rescues the deficit in dopamine neurotransmission in a mouse model of DYT1 dystonia. Neurobiology of Disease, 2019, 125, 115-122.	4.4	28
103	Stress, caffeine and ethanol trigger transient neurological dysfunction through shared mechanisms in a mouse calcium channelopathy. Neurobiology of Disease, 2013, 50, 151-159.	4.4	27
104	Temporal profile of improvement of tardive dystonia after globus pallidus deep brain stimulation. Parkinsonism and Related Disorders, 2015, 21, 116-119.	2.2	27
105	Medical and Surgical Treatments for Dystonia. Neurologic Clinics, 2020, 38, 325-348.	1.8	27
106	Pallidal Activity in Cervical Dystonia with and Without Head Tremor. Cerebellum, 2020, 19, 409-418.	2.5	27
107	Quality of life in isolated dystonia: non-motor manifestations matter. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 622-628.	1.9	27
108	Localization of hypoxanthine-guanine phosphoribosyltransferase mRNA in the mouse brain by in situ hybridization. Molecular and Cellular Neurosciences, 1992, 3, 64-78.	2.2	24

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109	Self-Biting Induced by Activation of L-Type Calcium Channels in Mice: Serotonergic Influences. Developmental Neuroscience, 2002, 24, 322-327.	2.0	24
110	Convergent mechanisms in etiologically-diverse dystonias. Expert Opinion on Therapeutic Targets, 2011, 15, 1387-1403.	3.4	24
111	Personalized Chemotherapy Profiling Using Cancer Cell Lines from Selectable Mice. Clinical Cancer Research, 2013, 19, 1139-1146.	7.0	24
112	Self-Biting Induced by Activation of L-Type Calcium Channels in Mice: Dopaminergic Influences. Developmental Neuroscience, 2003, 25, 20-25.	2.0	23
113	Nifedipine Suppresses Self-Injurious Behaviors in Animals. Developmental Neuroscience, 2007, 29, 241-250.	2.0	23
114	A double-blind, placebo-controlled, crossover trial of the selective dopamine D1 receptor antagonist ecopipam in patients with Lesch-Nyhan disease. Molecular Genetics and Metabolism, 2016, 118, 160-166.	1.1	23
115	Compositional complexity of rods and rings. Molecular Biology of the Cell, 2018, 29, 2303-2316.	2.1	23
116	Sudden death in Lesch-Nyhan disease. Developmental Medicine and Child Neurology, 2006, 48, 923.	2.1	23
117	Neuroanatomical substrates for paroxysmal dyskinesia in lethargic mice. Neurobiology of Disease, 2007, 27, 249-257.	4.4	22
118	Kinematic and electromyographic tools for characterizing movement disorders in mice. Movement Disorders, 2010, 25, 265-274.	3.9	22
119	Amphetamine-induced behavioral phenotype in a hypoxanthine^guanine phosphoribosyltransferase-deficient mouse model of Lesch-Nyhan syndrome.. Behavioral Neuroscience, 1991, 105, 1004-1012.	1.2	22
120	Mechanisms for phenotypic variation in Leschâ€“Nyhan disease and its variants. Human Genetics, 2011, 129, 71-78.	3.8	21
121	Secured web-based video repository for multicenter studies. Parkinsonism and Related Disorders, 2015, 21, 366-371.	2.2	21
122	The Dystonias. CONTINUUM Lifelong Learning in Neurology, 2019, 25, 976-1000.	0.8	21
123	Tetrahydrobiopterin deficiency and dopamine loss in a genetic mouse model of Lesch-Nyhan disease. Journal of Inherited Metabolic Disease, 2004, 27, 165-178.	3.6	20
124	The Role of Dopamine Receptors in the Neurobehavioral Syndrome Provoked by Activation of L-Type Calcium Channels in Rodents. Developmental Neuroscience, 2006, 28, 505-517.	2.0	20
125	Extreme task specificity in writer's cramp. Movement Disorders, 2011, 26, 2107-2109.	3.9	20
126	Objective, computerized video-based rating of blepharospasm severity. Neurology, 2016, 87, 2146-2153.	1.1	20

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127	Dopamine Receptor Agonist Treatment of Idiopathic Dystonia: A Reappraisal in Humans and Mice. Journal of Pharmacology and Experimental Therapeutics, 2018, 365, 20-26.	2.5	20
128	Oromandibular Dystonia: A Clinical Examination of 2,020 Cases. Frontiers in Neurology, 2021, 12, 700714.	2.4	20
129	Consequences of Delayed Dental Extraction in Leschâ€Nyhan Disease. Movement Disorders Clinical Practice, 2014, 1, 225-229.	1.5	19
130	Soft Nanomembrane Sensors and Flexible Hybrid Bioelectronics for Wireless Quantification of Blepharospasm. IEEE Transactions on Biomedical Engineering, 2020, 67, 3094-3100.	4.2	19
131	Expression of c-fos in the Brain after Activation of L-Type Calcium Channels. Developmental Neuroscience, 2003, 25, 403-411.	2.0	18
132	Gene therapy and the brain. British Medical Bulletin, 1995, 51, 138-148.	6.9	17
133	Globus pallidus deep brain stimulation for adult-onset axial dystonia. Parkinsonism and Related Disorders, 2014, 20, 1279-1282.	2.2	17
134	Why are voluntary head movements in cervical dystonia slow?. Parkinsonism and Related Disorders, 2015, 21, 561-566.	2.2	17
135	Cervical dystonia and substance abuse. Journal of Neurology, 2018, 265, 970-975.	3.6	17
136	Pharmacologic thresholds for self-injurious behavior in a genetic mouse model of Leschâ€“Nyhan disease. Pharmacology Biochemistry and Behavior, 2002, 73, 583-592.	2.9	16
137	Clinical subtypes of anterocollis in parkinsonian syndromes. Journal of the Neurological Sciences, 2012, 315, 100-103.	0.6	16
138	The Dystonia Coalition: A Multicenter Network for Clinical and Translational Studies. Frontiers in Neurology, 2021, 12, 660909.	2.4	16
139	Designing Clinical Trials for Dystonia. Neurotherapeutics, 2014, 11, 117-127.	4.4	15
140	Physiology of midbrain head movement neurons in cervical dystonia. Movement Disorders, 2017, 32, 904-912.	3.9	15
141	Predictors of alcohol responsiveness in dystonia. Neurology, 2018, 91, e2020-e2026.	1.1	15
142	Dystonia. Neurology: Clinical Practice, 2015, 5, 232-240.	1.6	14
143	Neural Substrates for Head Movements in Humans: A Functional Magnetic Resonance Imaging Study. Journal of Neuroscience, 2015, 35, 9163-9172.	3.6	14
144	A clinical trial of safety and tolerability for the selective dopamine D1 receptor antagonist ecopipam in patients with Lesch-Nyhan disease. Molecular Genetics and Metabolism, 2016, 117, 401-406.	1.1	14

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145	Potential mechanisms for low uric acid in Parkinson disease. Journal of Neural Transmission, 2016, 123, 365-370.	2.8	14
146	Macrocytic anemia in Leschâ€“Nyhan disease and its variants. Genetics in Medicine, 2019, 21, 353-360.	2.4	14
147	Physiological levels of folic acid reveal purine alterations in Lesch-Nyhan disease. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 12071-12079.	7.1	13
148	Phenotypic variation among seven members of one family with deficiency of hypoxanthineâ€“guanine phosphoribosyltransferase. Molecular Genetics and Metabolism, 2013, 110, 268-274.	1.1	12
149	Dystonia and cerebellar degeneration in the leaner mouse mutant. Brain Research, 2015, 1611, 56-64.	2.2	12
150	Soft Materialâ€Enabled, Active Wireless, Thinâ€Film Bioelectronics for Quantitative Diagnostics of Cervical Dystonia. Advanced Materials Technologies, 2019, 4, 1900458.	5.8	12
151	Gene expression and mRNA editing of serotonin receptor 2C in brains of HPRT gene knock-out mice, an animal model of Lesch-Nyhan disease. Journal of Clinical Neuroscience, 2009, 16, 1061-1063.	1.5	11
152	Parkinsonism without dopamine neuron degeneration in aged <scp>l</scp>â€dopaâ€responsive dystonia knockin mice. Movement Disorders, 2017, 32, 1694-1700.	3.9	11
153	Impaired Saccade Adaptation in Tremor-Dominant Cervical Dystoniaâ€”Evidence for Maladaptive Cerebellum. Cerebellum, 2021, 20, 678-686.	2.5	11
154	Clinical Features and Evolution of Blepharospasm: A Multicenter International Cohort and Systematic Literature Review. , 0, 1, .		11
155	Consequences of impaired purine recycling on the proteome in a cellular model of Leschâ€“Nyhan disease. Molecular Genetics and Metabolism, 2015, 114, 570-579.	1.1	10
156	Pilot Single-Blind Trial of AbobotulinumtoxinA in Oromandibular Dystonia. Neurotherapeutics, 2018, 15, 452-458.	4.4	10
157	Diagnostic and clinical experience of patients with pantothenate kinase-associated neurodegeneration. Orphanet Journal of Rare Diseases, 2019, 14, 174.	2.7	10
158	Cerebellar Dysfunction as a Source of Dystonic Phenotypes in Mice. Cerebellum, 2023, 22, 719-729.	2.5	10
159	A strategy for managing flu-like symptoms after botulinum toxin injections. Journal of Neurology, 2018, 265, 1932-1933.	3.6	9
160	A Scale to Assess Activities of Daily Living in Pantothenate Kinaseâ€Associated Neurodegeneration. Movement Disorders Clinical Practice, 2019, 6, 139-149.	1.5	9
161	Cell-intrinsic effects of TorsinA(Î”E) disrupt dopamine release in a mouse model of TOR1A dystonia. Neurobiology of Disease, 2021, 155, 105369.	4.4	9
162	Transcriptomic Approach to Lesch-Nyhan Disease. Nucleosides, Nucleotides and Nucleic Acids, 2014, 33, 208-217.	1.1	8

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163	Gene therapy for disorders of the nervous system. Trends in Biotechnology, 1993, 11, 192-197.	9.3	7
164	Do clinical features of Leschâ€Nyhan disease correlate more closely with hypoxanthine or guanine recycling?. Journal of Inherited Metabolic Disease, 2016, 39, 85-91.	3.6	7
165	New approaches to discovering drugs that treat dystonia. Expert Opinion on Drug Discovery, 2019, 14, 893-900.	5.0	7
166	Neuronal Activity of Pallidal Versus Cerebellar Receiving Thalamus in Patients with Cervical Dystonia. Cerebellum, 2021, 20, 151-159.	2.5	7
167	Adapting to post-COVID19 research in Parkinson's disease: Lessons from a multinational experience. Parkinsonism and Related Disorders, 2021, 82, 146-149.	2.2	7
168	Induced pluripotent stem cells from subjects with Lesch-Nyhan disease. Scientific Reports, 2021, 11, 8523.	3.3	7
169	Neuropathology of blepharospasm. Experimental Neurology, 2021, 346, 113855.	4.1	7
170	Bone marrow transplantation does not ameliorate the neurologic symptoms in mice deficient in hypoxanthine guanine phosphoribosyl transferase (HPRT). Metabolic Brain Disease, 1999, 14, 57-65.	2.9	6
171	NEURONAL VOLTAGE-GATED CALCIUM CHANNELS: BRIEF OVERVIEW OF THEIR FUNCTION AND CLINICAL IMPLICATIONS IN NEUROLOGY. Neurology, 2010, 75, 937-938.	1.1	6
172	Ocular Palatal Tremor Plus Dystonia: New Syndromic Association. Movement Disorders Clinical Practice, 2015, 2, 267-270.	1.5	6
173	A metabolomic study of cervical dystonia. Parkinsonism and Related Disorders, 2021, 82, 98-103.	2.2	6
174	The Motor Disorder of Classic Leschâ€Nyhan Disease. Nucleosides, Nucleotides and Nucleic Acids, 2004, 23, 1161-1164.	1.1	5
175	Behavioral aspects of Lesch-Nyhan disease and its variants. Developmental Medicine and Child Neurology, 2007, 47, 673-677.	2.1	5
176	â€œComplexâ€•dystonia is not a category in the new 2013 consensus classification. Movement Disorders, 2016, 31, 1758-1759.	3.9	5
177	Blockade of M4 muscarinic receptors on striatal cholinergic interneurons normalizes striatal dopamine release in a mouse model of TOR1A dystonia. Neurobiology of Disease, 2022, 168, 105699.	4.4	5
178	Differential expression of striatal proteins in a mouse model of DOPA-responsive dystonia reveals shared mechanisms among dystonic disorders. Molecular Genetics and Metabolism, 2021, 133, 352-361.	1.1	4
179	Locus Pocus. Movement Disorders, 2016, 31, 1759-1760.	3.9	3
180	Expert Opinion vs Patient Perspective in Treatment ofÂRareÂDisorders: Tooth Removal in Lesch-Nyhan Disease asÂanÂExample. JIMD Reports, 2018, 41, 25-27.	1.5	3

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181	Neurological research & training after the easing of lockdown in countries impacted by COVID-19. Journal of the Neurological Sciences, 2020, 418, 117105.	0.6	3
182	The emerging neurological spectrum of AARS2-associated disorders. Parkinsonism and Related Disorders, 2021, 93, 50-54.	2.2	3
183	The dystonias: Past, present, and future. Movement Disorders, 2013, 28, 849-850.	3.9	2
184	Microstructural white matter abnormalities in Leschâ€“Nyhan disease. European Journal of Neuroscience, 2022, 55, 264-276.	2.6	2
185	Leschâ€“Nyhan disease with no HPRT1 gene mutation?. Revista Clinica Espanola, 2014, 214, 459-460.	0.6	1
186	Combined occurrence of deleterious TOR1A and ANO3 variants in isolated generalized dystonia. Parkinsonism and Related Disorders, 2020, 73, 55-56.	2.2	1
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