Li Cao

List of Publications by Year
in descending order

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83	1,769	361296	330025
papers	citations	20 h-index	37 g-index

89	89	89	1824
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Identification of PRRT2 as the causative gene of paroxysmal kinesigenic dyskinesias. Brain, 2011, 134, 3493-3501.	3.7	263
2	Long-read sequencing identified repeat expansions in the 5â€²UTR of the NOTCH2NLC gene from Chinese patients with neuronal intranuclear inclusion disease. Journal of Medical Genetics, 2019, 56, 758-764.	1.5	94
3	Expansion of GGC Repeat in GIPC1 Is Associated with Oculopharyngodistal Myopathy. American Journal of Human Genetics, 2020, 106, 793-804.	2.6	90
4	The GGC repeat expansion inNOTCH2NLCis associated with oculopharyngodistal myopathy type 3. Brain, 2021, 144, 1819-1832.	3.7	81
5	Translation of GGC repeat expansions into a toxic polyglycine protein in NIID defines a novel class of human genetic disorders: The polyG diseases. Neuron, 2021, 109, 1825-1835.e5.	3.8	79
6	CGG expansion in NOTCH2NLC is associated with oculopharyngodistal myopathy with neurological manifestations. Acta Neuropathologica Communications, 2020, 8, 204.	2.4	76
7	Paroxysmal kinesigenic dyskinesia. Neurology, 2015, 85, 1546-1553.	1.5	72
8	The prevalence of LRRK2 Gly2385Arg variant in Chinese Han population with Parkinson's disease. Movement Disorders, 2007, 22, 2439-2443.	2.2	62
9	Identification of a novel PRRT2 mutation in patients with paroxysmal kinesigenic dyskinesias and c.649dupC as a mutation hot-spot. Parkinsonism and Related Disorders, 2012, 18, 704-706.	1.1	51
10	A common region of allelic loss on chromosome region 3p25.3â€“26.3 in nasopharyngeal carcinoma. Genes Chromosomes and Cancer, 1998, 23, 21-25.	1.5	50
11	Familial paroxysmal kinesigenic dyskinesia is associated with mutations in the KCNA1 gene. Human Molecular Genetics, 2018, 27, 625-637.	1.4	43
12	Clinical and pathological features in adult-onset NIID patients with cortical enhancement. Journal of Neurology, 2020, 267, 3187-3198.	1.8	43
13	Prolineâ€rich transmembrane protein 2â€“negative paroxysmal kinesigenic dyskinesia: Clinical and genetic analyses of 163 patients. Movement Disorders, 2018, 33, 459-467.	2.2	41
14	(CAG) _n loci as genetic modifiers of age-at-onset in patients with Machado-Joseph disease from mainland China. Brain, 2016, 139, e41-e41.	3.7	37
15	The CGG repeat expansion in RILPL1 is associated with oculopharyngodistal myopathy type 4. American Journal of Human Genetics, 2022, 109, 533-541.	2.6	35
16	The copy number of Epstein-Barr virus latent genome correlates with the oncogenicity by the activation level of LMP1 and NF-ÎºB. Oncotarget, 2015, 6, 41033-41044.	0.8	34
17	Severe sensory neuropathy in patients with adult-onset multiple acyl-CoA dehydrogenase deficiency. Neuromuscular Disorders, 2016, 26, 170-175.	0.3	29
18	The Phenotypic and Genetic Spectrum of Paroxysmal Kinesigenic Dyskinesia in China. Movement Disorders, 2020, 35, 1428-1437.	2.2	28

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19	Clinical and Muscle Imaging Findings in 14 Mainland Chinese Patients with Oculopharyngodistal Myopathy. PLoS ONE, 2015, 10, e0128629.	1.1	26
20	The Whole Exome Sequencing Clarifies the Genotype- Phenotype Correlations in Patients with Early-Onset Dementia. , 2018, 9, 696.		26
21	PRRT2 truncated mutations lead to nonsense-mediated mRNA decay in Paroxysmal Kinesigenic Dyskinesia. Parkinsonism and Related Disorders, 2014, 20, 1399-1404.	1.1	23
22	Clinicopathologic characterization and abnormal autophagy of CSF1R-related leukoencephalopathy. Translational Neurodegeneration, 2019, 8, 32.	3.6	21
23	Myotonia congenita: novel mutations in CLCN1 gene. Channels, 2015, 9, 292-298.	1.5	20
24	<scp>TMEM151A</scp> Variants Cause Paroxysmal Kinesigenic Dyskinesia: A Largeâ€Sample Study. Movement Disorders, 2022, 37, 545-552.	2.2	20
25	Recommendations for the diagnosis and treatment of paroxysmal kinesigenic dyskinesia: an expert consensus in China. Translational Neurodegeneration, 2021, 10, 7.	3.6	19
26	GGC repeat expansions in NOTCH2NLC causing a phenotype of distal motor neuropathy and myopathy. Annals of Clinical and Translational Neurology, 2021, 8, 1330-1342.	1.7	18
27	The polyG diseases: a new disease entity. Acta Neuropathologica Communications, 2022, 10, .	2.4	18
28	The quality of life in papillary thyroid microcarcinoma patients undergoing lobectomy or total thyroidectomy: A crossâ€sectional study. Cancer Medicine, 2021, 10, 1989-2002.	1.3	16
29	Limb-girdle congenital myasthenic syndrome in a Chinese family with novel mutations in MUSK gene and literature review. Clinical Neurology and Neurosurgery, 2016, 150, 41-45.	0.6	15
30	Expanding the clinical spectrum of adult-onset neuronal intranuclear inclusion disease. Acta Neurologica Belgica, 2022, 122, 647-658.	0.5	15
31	Complicated paroxysmal kinesigenic dyskinesia associated with SACS mutations. Annals of Translational Medicine, 2020, 8, 8-8.	0.7	14
32	Study on the safety and effectiveness of drug-coated balloons in patients with acute myocardial infarction. Journal of Cardiothoracic Surgery, 2021, 16, 178.	0.4	14
33	Progressive myoclonus epilepsy without renal failure in a Chinese family with a novel mutation in SCARB2 gene and literature review. Seizure: the Journal of the British Epilepsy Association, 2018, 57, 80-86.	0.9	13
34	Primary familial brain calcification presenting as paroxysmal kinesigenic dyskinesia: Genetic and functional analyses. Neuroscience Letters, 2020, 714, 134543.	1.0	13
35	ATL3 gene mutation in a Chinese family with hereditary sensory neuropathy type 1F. Journal of the Peripheral Nervous System, 2019, 24, 150-155.	1.4	12
36	GGC Repeat Expansion in the NOTCH2NLC Gene Is Associated With a Phenotype of Predominant Motorâ€“Sensory and Autonomic Neuropathy. Frontiers in Genetics, 2021, 12, 694790.	1.1	12

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37	Infantile spinal muscular atrophy with respiratory distress type I presenting without respiratory involvement: Novel mutations and review of the literature. Brain and Development, 2016, 38, 685-689.	0.6	11
38	Altered structural and functional connectivity in CSF1R-related leukoencephalopathy. Brain Imaging and Behavior, 2020, 15, 1655-1666.	1.1	11
39	Novel ATM mutations with ataxia-telangiectasia. Neuroscience Letters, 2016, 611, 112-115.	1.0	10
40	Ubiquitin-related network underlain by (CAG)n loci modulate age at onset in Machado-Joseph disease. Brain, 2017, 140, e25-e25.	3.7	10
41	Novel mutations in the SPAST gene cause hereditary spastic paraplegia. Parkinsonism and Related Disorders, 2019, 69, 125-133.	1.1	10
42	Depression, Anxiety, and Quality of Life in Paroxysmal Kinesigenic Dyskinesia Patients. Chinese Medical Journal, 2017, 130, 2088-2094.	0.9	9
43	Lysosomal degradation of <scp>GMPPB</scp> is associated with limbâ€girdle muscular dystrophy type 2T. Annals of Clinical and Translational Neurology, 2019, 6, 1062-1071.	1.7	9
44	Advances in hyperekplexia and other startle syndromes. Neurological Sciences, 2021, 42, 4095-4107.	0.9	9
45	Detailed deletion mapping of chromosome 9p21â€“22 in nasopharyngeal carcinoma. Chinese Journal of Cancer Research: Official Journal of China Anti-Cancer Association, Beijing Institute for Cancer Research, 2000, 12, 161-164.	0.7	8
46	Case report: A Chinese child with Andersenâ€“Tawil syndrome due to a de novo KCNJ2 mutation. Journal of the Neurological Sciences, 2015, 352, 105-106.	0.3	8
47	Ataxia with novel compound heterozygous PEX10 mutations and a literature review of PEX10-related peroxisome biogenesis disorders. Clinical Neurology and Neurosurgery, 2019, 177, 92-96.	0.6	7
48	Congenital disorder of glycosylation type 1T with a novel truncated homozygous mutation in PGM1 gene and literature review. Neuromuscular Disorders, 2019, 29, 282-289.	0.3	7
49	New phenotype of DCTN1â€related spectrum: earlyâ€onset dHMN plus congenital foot deformity. Annals of Clinical and Translational Neurology, 2020, 7, 200-209.	1.7	7
50	Neurodevelopmental disorder caused by a truncating de novo variant of IRF2BPL. Seizure: the Journal of the British Epilepsy Association, 2021, 84, 47-52.	0.9	7
51	Variants in LAMC3 Causes Occipital Cortical Malformation. Frontiers in Genetics, 2021, 12, 616761.	1.1	7
52	Characteristics of ocular findings of patients with neuronal intranuclear inclusion disease. Neurological Sciences, 2022, 43, 3231-3237.	0.9	7
53	Novel mutations in the CYP7B1 gene cause hereditary spastic paraplegia. Movement Disorders, 2011, 26, 1354-1356.	2.2	6
54	Human-robot Interaction Oriented Human-in-the-loop Real-time Motion Imitation on a Humanoid Tri-Co Robot. , 2018, , .		6

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55	Novel Mutations in SCN4A Gene Cause Myotonia Congenita with Scoliosis. Chinese Medical Journal, 2018, 131, 477-479.	0.9	6
56	Novel PANK2 Mutations in Patients With Pantothenate Kinase-Associated Neurodegeneration and the Genotypeâ€“Phenotype Correlation. Frontiers in Aging Neuroscience, 2022, 14, 848919.	1.7	6
57	Novel compound heterozygous mutations in a GNE myopathy with congenital thrombocytopenia: A case report and literature review. Clinical Case Reports (discontinued), 2022, 10, e05659.	0.2	6
58	Thigh MRI in antisynthetase syndrome, and comparisons with dermatomyositis and immune-mediated necrotizing myopathy. Rheumatology, 2022, 62, 310-320.	0.9	6
59	Teaching Video NeuroImages: Cautious walking gait in siblings with hereditary hyperekplexia. Neurology, 2019, 92, e2068-e2069.	1.5	5
60	Sural nerve pathology inTFGâ€associated motor neuron disease with sensory neuropathy. Neuropathology, 2019, 39, 194-199.	0.7	5
61	Mechanisms and Pharmacotherapy for Ethanol-Responsive Movement Disorders. Frontiers in Neurology, 2020, 11, 892.	1.1	5
62	Clinical and genetic analyses of 150 patients with paroxysmal kinesigenic dyskinesia. Journal of Neurology, 2022, 269, 4717-4728.	1.8	5
63	c.1263+1G>A Is a Latent Hotspot for CYP27A1 Mutations in Chinese Patients With Cerebrotendinous Xanthomatosis. Frontiers in Genetics, 2020, 11, 682.	1.1	4
64	Case Report: Neuronal Intranuclear Inclusion Disease With Oromandibular Dystonia Onset. Frontiers in Neurology, 2021, 12, 618595.	1.1	4
65	Novel IBA57 mutations in two chinese patients and literature review of multiple mitochondrial dysfunction syndrome. Metabolic Brain Disease, 2022, 37, 311-317.	1.4	4
66	Coverage of tuberculosis and diabetes mellitus screening among household contacts of tuberculosis patients: a household-based cross-sectional survey from Southern Thailand. BMC Public Health, 2020, 20, 957.	1.2	3
67	The Prognostic Value of Serum Uric Acid in Hospitalized Patients with Acute Cerebral Infarction. Disease Markers, 2021, 2021, 1-8.	0.6	3
68	Excessive Startle with Novel GLRA1 Mutations in 4 Chinese Patients and a Literature Review of		

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73	A report of two cases of bulbospinal form Alexander disease and preliminary exploration of the disease. Molecular Medicine Reports, 2021, 24, .	1.1	2
74	Health services utilization of Chinese patients with Huntingtonâ€™s disease: a cross-sectional study. BMC Health Services Research, 2021, 21, 806.	0.9	2
75	Generation of an human induced pluripotent stem cell JTUi007-A from a patient with CSF1R-related leukoencephalopathy carrying heterozygous p.Ile794Thr mutation in CSF1R gene. Stem Cell Research, 2021, 57, 102593.	0.3	2
76	Altered Local Brain Amplitude of Fluctuations in Patients With Myotonic Dystrophy Type 1. Frontiers in Aging Neuroscience, 2021, 13, 790632.	1.7	1
77	Altered intrinsic brain activity in patients with CSF1R-related leukoencephalopathy. Brain Imaging and Behavior, 2022, 16, 1842-1853.	1.1	1
78	New phenotype of <scp>RTN2</scp>â€related spectrum: Complicated form of spastic paraplegiaâ€12. Annals of Clinical and Translational Neurology, 2022, 9, 1108-1115.	1.7	1
79	Autosomal dominant hypocalcemia with a novel CASR mutation: a case study and literature review. Journal of International Medical Research, 2022, 50, 030006052211104.	0.4	1
80	The DNase-1 sensitive regions in genomes of Burkittâ€™s lymphoma cells. Chinese Journal of Cancer Research: Official Journal of China Anti-Cancer Association, Beijing Institute for Cancer Research, 1993, 5, 245-251.	0.7	0
81	Expression of nitroreductase gene NOR1 in E.Coli and the preparation of antiserum. Chinese Journal of Cancer Research: Official Journal of China Anti-Cancer Association, Beijing Institute for Cancer Research, 2004, 16, 11-14.	0.7	0
82	Identification of disregulated cell cycle pathway in nasopharyngeal carcinoma by gene set enrichment analysis. Cell Biology International, 2008, 32, S39-S39.	1.4	0
83	Novel mutation of the PRRT2 gene in two cases of paroxysmal kinesigenic dyskinesia: Two case reports. Biomedical Reports, 2020, 12, 309-312.	0.9	0