List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Identification of PRRT2 as the causative gene of paroxysmal kinesigenic dyskinesias. Brain, 2011, 134, 3493-3501.	7.6	263
2	Expansion of Human-Specific GGC Repeat in Neuronal Intranuclear Inclusion Disease-Related Disorders. American Journal of Human Genetics, 2019, 105, 166-176.	6.2	212
3	Expansion of GGC repeat in the human-specific NOTCH2NLC gene is associated with essential tremor. Brain, 2020, 143, 222-233.	7.6	139
4	The role of genetics in Parkinson's disease: a large cohort study in Chinese mainland population. Brain, 2020, 143, 2220-2234.	7.6	97
5	Primary erythromelalgia: a review. Orphanet Journal of Rare Diseases, 2015, 10, 127.	2.7	90
6	<i>PLA2G6</i> gene mutation in autosomal recessive early-onset parkinsonism in a Chinese cohort. Neurology, 2011, 77, 75-81.	1.1	87
7	Long-read sequencing identified intronic repeat expansions in <i>SAMD12</i> from Chinese pedigrees affected with familial cortical myoclonic tremor with epilepsy. Journal of Medical Genetics, 2019, 56, 265-270.	3.2	82
8	Coding mutations inNUS1contribute to Parkinson's disease. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 11567-11572.	7.1	78
9	Polygenic determinants of Parkinson's disease in a Chinese population. Neurobiology of Aging, 2015, 36, 1765.e1-1765.e6.	3.1	73
10	Mutation analysis of <i>Parkin</i> , <i>PINK1</i> , <i>DJâ€1</i> and <i>ATP13A2</i> genes in Chinese patients with autosomal recessive earlyâ€onset Parkinsonism. Movement Disorders, 2008, 23, 2074-2079.	3.9	61
11	Identification of GGC repeat expansion in the <i>NOTCH2NLC</i> gene in amyotrophic lateral sclerosis. Neurology, 2020, 95, e3394-e3405.	1.1	59
12	Investigation of Gene Regulatory Networks Associated with Autism Spectrum Disorder Based on MiRNA Expression in China. PLoS ONE, 2015, 10, e0129052.	2.5	50
13	Analysis of SCA2 and SCA3/MJD repeats in Parkinson's disease in mainland China: Genetic, clinical, and positron emission tomography findings. Movement Disorders, 2009, 24, 2007-2011.	3.9	43
14	Familial paroxysmal kinesigenic dyskinesia is associated with mutations in the KCNA1 gene. Human Molecular Genetics, 2018, 27, 625-637.	2.9	43
15	Alterations of the Gut Microbiota in Multiple System Atrophy Patients. Frontiers in Neuroscience, 2019, 13, 1102.	2.8	42
16	Biallelic Intronic <scp>AAGGG</scp> Expansion of <scp><i>RFC1</i></scp> is Related to Multiple System Atrophy. Annals of Neurology, 2020, 88, 1132-1143.	5.3	41
17	miRâ€25 alleviates polyQâ€mediated cytotoxicity by silencing <i>ATXN3</i> . FEBS Letters, 2014, 588, 4791-4798.	2.8	37
18	(CAG) _n loci as genetic modifiers of age-at-onset in patients with Machado-Joseph disease from mainland China. Brain, 2016, 139, e41-e41.	7.6	37

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19	Regional Brain and Spinal Cord Volume Loss in Spinocerebellar Ataxia Type 3. Movement Disorders, 2021, 36, 2273-2281.	3.9	37
20	Updated frequency analysis of spinocerebellar ataxia in China. Brain, 2018, 141, e22-e22.	7.6	33
21	GCH1 variants contribute to the risk and earlier age-at-onset of Parkinson's disease: a two-cohort case-control study. Translational Neurodegeneration, 2020, 9, 31.	8.0	30
22	The APOE ε2 allele may decrease the age at onset in patients with spinocerebellar ataxia type 3 or Machado-Joseph disease from the Chinese Han population. Neurobiology of Aging, 2014, 35, 2179.e15-2179.e18.	3.1	25
23	Identification of a potential exosomal biomarker in spinocerebellar ataxia Type 3/Machado–Joseph disease. Epigenomics, 2019, 11, 1037-1056.	2.1	23
24	Rare GCH1 heterozygous variants contributing to Parkinson's disease. Brain, 2017, 140, e41-e41.	7.6	21
25	Gene-Related Cerebellar Neurodegeneration in SCA3/MJD: A Case-Controlled Imaging-Genetic Study. Frontiers in Neurology, 2019, 10, 1025.	2.4	21
26	Paving the Way Toward Meaningful Trials in Ataxias: An Ataxia Global Initiative Perspective. Movement Disorders, 2022, 37, 1125-1130.	3.9	21
27	Huntingtin gene <scp>CAG</scp> repeat numbers in <scp>C</scp> hinese patients with <scp>H</scp> untington's disease and controls. European Journal of Neurology, 2014, 21, 637-642.	3.3	20
28	Identification of a de novo DYNC1H1 mutation via WES according to published guidelines. Scientific Reports, 2016, 6, 20423.	3.3	20
29	Genetic modifiers of age-at-onset in polyglutamine diseases. Ageing Research Reviews, 2018, 48, 99-108.	10.9	20
30	<scp><i>TMEM151A</i></scp> Variants Cause Paroxysmal Kinesigenic Dyskinesia: A Large‣ample Study. Movement Disorders, 2022, 37, 545-552.	3.9	20
31	Using next-generation sequencing as a genetic diagnostic tool in rare autosomal recessive neurologic Mendelian disorders. Neurobiology of Aging, 2013, 34, 2442.e11-2442.e17.	3.1	19
32	Two Novel SNPs in ATXN3 3' UTR May Decrease Age at Onset of SCA3/MJD in Chinese Patients. PLoS ONE, 2015, 10, e0117488.	2.5	19
33	Association of serum neurofilament light and disease severity in patients with spinocerebellar ataxia type 3. Neurology, 2020, 95, e2977-e2987.	1.1	19
34	Recommendations for the diagnosis and treatment of paroxysmal kinesigenic dyskinesia: an expert consensus in China. Translational Neurodegeneration, 2021, 10, 7.	8.0	19
35	Genotype and phenotype distribution of 435 patients with Charcot–Marie–Tooth disease from central south China. European Journal of Neurology, 2021, 28, 3774-3783.	3.3	19
36	Alteration of methylation status in the ATXN3 gene promoter region is linked to the SCA3/MJD. Neurobiology of Aging, 2017, 53, 192.e5-192.e10.	3.1	18

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37	MR Imaging of SCA3/MJD. Frontiers in Neuroscience, 2020, 14, 749.	2.8	18
38	Is the High Frequency of Machado-Joseph Disease in China Due to New Mutational Origins?. Frontiers in Genetics, 2018, 9, 740.	2.3	17
39	A novel mutation in <i>MYORG</i> causes primary familial brain calcification with central neuropathic pain. Clinical Genetics, 2019, 95, 433-435.	2.0	17
40	Mutation analysis of hereditary multiple exostoses in the Chinese. Human Genetics, 1999, 105, 45-50.	3.8	16
41	<i>ATXN2</i> polymorphism modulates age at onset in Machado-Joseph disease. Brain, 2016, 139, aww176.	7.6	16
42	Association of <i>TNF-α</i> rs1799964 and <i>IL-1β</i> rs16944 polymorphisms with multiple system atrophy in Chinese Han population. International Journal of Neuroscience, 2018, 128, 761-764.	1.6	16
43	CRISPR/Cas9 mediated gene correction ameliorates abnormal phenotypes in spinocerebellar ataxia type 3 patient-derived induced pluripotent stem cells. Translational Psychiatry, 2021, 11, 479.	4.8	15
44	Identifying SYNE1 Ataxia With Novel Mutations in a Chinese Population. Frontiers in Neurology, 2018, 9, 1111.	2.4	14
45	Genetic and clinical analysis of spinocerebellar ataxia type 36 in Mainland China. Clinical Genetics, 2016, 90, 141-148.	2.0	13
46	Roles of Post-translational Modifications in Spinocerebellar Ataxias. Frontiers in Cellular Neuroscience, 2018, 12, 290.	3.7	13
47	Birt-Hogg-Dubé syndrome in two Chinese families with mutations in the FLCN gene. BMC Medical Genetics, 2018, 19, 14.	2.1	13
48	Spinocerebellar ataxia type 27 (SCA27) is an uncommon cause of dominant ataxia among Chinese Han population. Neuroscience Letters, 2012, 520, 16-19.	2.1	12
49	Novel mutations in ADSL for Adenylosuccinate Lyase Deficiency identified by the combination of Trio-WES and constantly updated guidelines. Scientific Reports, 2017, 7, 1625.	3.3	12
50	Analysis of the GGGGCC Repeat Expansions of the C9orf72 Gene in SCA3/MJD Patients from China. PLoS ONE, 2015, 10, e0130336.	2.5	11
51	Analysis of (CAG)n expansion in ATXN1, ATXN2 and ATXN3 in Chinese patients with multiple system atrophy. Scientific Reports, 2018, 8, 3889.	3.3	11
52	(<scp>CAG</scp>) _{<i>n</i>} loci as genetic modifiers of age at onset in patients with spinocerebellar ataxia type 1 from mainland China. European Journal of Neurology, 2019, 26, 1130-1136.	3.3	11
53	Prediction of the Age at Onset of Spinocerebellar Ataxia Type 3 with Machine Learning. Movement Disorders, 2021, 36, 216-224.	3.9	11
54	Cholecystokinin 1 receptor activation restores normal mTORC1 signaling and is protective to Purkinje cells of SCA mice. Cell Reports, 2021, 37, 109831.	6.4	11

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55	Ubiquitin-related network underlain by (CAG)n loci modulate age at onset in Machado-Joseph disease. Brain, 2017, 140, e25-e25.	7.6	10
56	High Serum GFAP Levels in SCA3/MJD May Not Correlate with Disease Progression. Cerebellum, 2015, 14, 677-681.	2.5	9
57	ATP10B variants in Parkinson's disease: a large cohort study in Chinese mainland population. Acta Neuropathologica, 2021, 141, 805-806.	7.7	8
58	Blood Neurofilament Light Chain in Genetic Ataxia: A Metaâ€Analysis. Movement Disorders, 2022, 37, 171-181.	3.9	8
59	Targeted Next-Generation Sequencing Revealed Novel Mutations in Chinese Ataxia Telangiectasia Patients: A Precision Medicine Perspective. PLoS ONE, 2015, 10, e0139738.	2.5	8
60	Genetic and clinical analyses of spinocerebellar ataxia type 8 in mainland China. Journal of Neurology, 2019, 266, 2979-2986.	3.6	7
61	New Model for Estimation of the Age at Onset in Spinocerebellar Ataxia Type 3. Neurology, 2021, 96, e2885-e2895.	1.1	7
62	Anxiety and depression in spinocerebellar ataxia patients during the COVID-19 pandemic in China: A cross-sectional study. Journal of Clinical Neuroscience, 2021, 88, 39-46.	1.5	7
63	<i>>PSAP</i> variants in Parkinson's disease: a large cohort study in Chinese mainland population. Brain, 2021, 144, e25-e25.	7.6	7
64	miRNA profiling in autism spectrum disorder in China. Genomics Data, 2015, 6, 108-109.	1.3	6
65	Generation of an induced pluripotent stem cell line (XHCSUi001-A) from urine cells of a patient with spinocerebellar ataxia type 3. Stem Cell Research, 2019, 40, 101555.	0.7	6
66	Generation of induced pluripotent stem cell line (CSUXHi002-A) from a patient with spinocerebellar ataxia type 1. Stem Cell Research, 2020, 45, 101816.	0.7	6
67	Central motor conduction time in spinocerebellar ataxia: a meta-analysis. Aging, 2020, 12, 25718-25729.	3.1	6
68	Cerebellar IncRNA Expression Profile Analysis of SCA3/MJD Mice. International Journal of Genomics, 2018, 2018, 1-6.	1.6	5
69	Investigation on modulation of DNA repair pathways in Chinese MJD patients. Neurobiology of Aging, 2018, 71, 267.e5-267.e6.	3.1	5
70	Clinical findings of autosomal-dominant striatal degeneration and PDE8B mutation screening in parkinsonism and related disorders. Parkinsonism and Related Disorders, 2019, 69, 94-98.	2.2	5
71	Polymorphisms in DNA methylation–related genes are linked to the phenotype of Machado-Joseph disease. Neurobiology of Aging, 2019, 75, 225.e1-225.e8.	3.1	5
72	Rare, pathogenic variants in LRP10 are associated with amyotrophic lateral sclerosis in patients from mainland China. Neurobiology of Aging, 2021, 97, 145.e17-145.e22.	3.1	5

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73	No genetic evidence for the involvement of GGC repeat expansions of the NOTCH2NLC gene in Chinese patients with multiple system atrophy. Neurobiology of Aging, 2021, 97, 144.e5-144.e7.	3.1	5
74	Human stem cell models of polyglutamine diseases: Sources for disease models and cell therapy. Experimental Neurology, 2021, 337, 113573.	4.1	5
75	<i>UQCRC1</i> variants in Parkinson's disease: a large cohort study in Chinese mainland population. Brain, 2021, 144, e54-e54.	7.6	5
76	Genetic etiology of a Chinese ataxia cohort: Expanding the mutational spectrum of hereditary ataxias. Parkinsonism and Related Disorders, 2021, 89, 120-127.	2.2	5
77	The Effects of Dracocephalum Heterophyllum Benth Flavonoid on Hypertrophic Cardiomyocytes Induced by Angiotensin II in Rats. Medical Science Monitor, 2018, 24, 6322-6330.	1.1	5
78	The progression rate of spinocerebellar ataxia type 3 varies with disease stage. Journal of Translational Medicine, 2022, 20, 226.	4.4	5
79	RNA Expression Profile and Potential Biomarkers in Patients With Spinocerebellar Ataxia Type 3 From Mainland China. Frontiers in Genetics, 2019, 10, 566.	2.3	4
80	Age is an important independent modifier of SCA3 phenotype severity. Neuroscience Letters, 2021, 741, 135510.	2.1	4
81	Mutation analysis of CAPN1 in Chinese populations with spastic paraplegia and related neurodegenerative diseases. Journal of the Neurological Sciences, 2020, 411, 116691.	0.6	4
82	Polyglutamine-expanded ataxin3 alter specific gene expressions through changing DNA methylation status in SCA3/MJD. Aging, 2021, 13, 3680-3698.	3.1	4
83	Profiling the Genome-Wide Landscape of Short Tandem Repeats by Long-Read Sequencing. Frontiers in Genetics, 2022, 13, .	2.3	4
84	Micro-structural white matter abnormalities and cognitive impairment in asymptomatic carotid plaque patients: A DTI study using TBSS analysis. Clinical Neurology and Neurosurgery, 2020, 197, 106096.	1.4	3
85	Generation of spinocerebellar ataxia type 3 patient-derived induced pluripotent stem cell line (CSUXHi005-A) from human urine epithelial cells. Stem Cell Research, 2021, 53, 102289.	0.7	3
86	Identification of the Largest SCA36 Pedigree in Asia: with Multimodel Neuroimaging Evaluation for the First Time. Cerebellum, 2022, 21, 358-367.	2,5	3
87	Myeleterosis in an ALPS5 patient with primary immune dysregulation syndrome. CNS Neuroscience and Therapeutics, 2020, 26, 773-775.	3.9	1
88	Profiling of mitochondrial genomes in SCA3/MJD patients from mainland China. Gene, 2020, 738, 144487.	2.2	1
89	Coffin-Siris syndrome in two chinese patients with novel pathogenic variants of ARID1A and SMARCA4. Genes and Genomics, 2022, , 1.	1.4	1
90	Noncoding RNAs and Base Modifications: Epigenomic Players Implicated in Neurological Disorders and Tumorigenesis. International Journal of Genomics, 2018, 2018, 1-2.	1.6	0

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91	Identification of novel mutations in TSC1 and TSC2 for tuberous sclerosis complex by targeted next-generation sequencing and ACMG guidelines. Child's Nervous System, 2020, 36, 1827-1830.	1.1	Ο
92	A Variant in Genes of the NPY System as Modifier Factor of Machado-Joseph Disease in the Chinese Population. Frontiers in Aging Neuroscience, 2022, 14, 822657.	3.4	0