

Zhi-Ying Wu

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148
papers

2,152
citations

21
h-index

40
g-index

165
ext. papers

2,707
ext. citations

5.6
avg, IF

5.09
L-index

#	Paper	IF	Citations
148	Exome sequencing identifies truncating mutations in PRRT2 that cause paroxysmal kinesigenic dyskinesia. <i>Nature Genetics</i> , 2011 , 43, 1252-5	36.3	339
147	Near-Infrared Upconversion Chemodosimeter for In Vivo Detection of Cu(2+) in Wilson Disease. <i>Advanced Materials</i> , 2016 , 28, 6625-30	24	89
146	Spinocerebellar ataxia: relationship between phenotype and genotype - a review. <i>Clinical Genetics</i> , 2016 , 90, 305-14	4	79
145	Mutation analysis and the correlation between genotype and phenotype of Arg778Leu mutation in chinese patients with Wilson disease. <i>Archives of Neurology</i> , 2001 , 58, 971-6		67
144	Molecular analysis of 51 unrelated pedigrees with late-onset multiple acyl-CoA dehydrogenation deficiency (MADD) in southern China confirmed the most common ETFDH mutation and high carrier frequency of c.250G>A. <i>Journal of Molecular Medicine</i> , 2011 , 89, 569-76	5.5	53
143	A striatal-enriched intronic GPCR modulates huntingtin levels and toxicity. <i>ELife</i> , 2015 , 4,	8.9	49
142	Spectrum and Classification of ATP7B Variants in a Large Cohort of Chinese Patients with Wilson Disease Guides Genetic Diagnosis. <i>Theranostics</i> , 2016 , 6, 638-49	12.1	49
141	PRRT2 deficiency induces paroxysmal kinesigenic dyskinesia by regulating synaptic transmission in cerebellum. <i>Cell Research</i> , 2018 , 28, 90-110	24.7	48
140	PRRT2 mutation correlated with phenotype of paroxysmal kinesigenic dyskinesia and drug response. <i>Neurology</i> , 2013 , 80, 1534-5	6.5	47
139	Mutation analysis of 218 Chinese patients with Wilson disease revealed no correlation between the canine copper toxicosis gene MURR1 and Wilson disease. <i>Journal of Molecular Medicine</i> , 2006 , 84, 438-42	5.5	44
138	Risk prediction for sporadic Alzheimer disease using genetic risk score in the Han Chinese population. <i>Oncotarget</i> , 2015 , 6, 36955-64	3.3	44
137	Genotype-phenotype correlations of amyotrophic lateral sclerosis. <i>Translational Neurodegeneration</i> , 2016 , 5, 3	10.3	43
136	Identify mutation in amyotrophic lateral sclerosis cases using HaloPlex target enrichment system. <i>Neurobiology of Aging</i> , 2014 , 35, 2881.e11-2881.e15	5.6	38
135	Neurofilament light chain is a promising serum biomarker in spinocerebellar ataxia type 3. <i>Molecular Neurodegeneration</i> , 2019 , 14, 39	19	35
134	High frequency of Machado-Joseph disease identified in southeastern Chinese kindreds with spinocerebellar ataxia. <i>BMC Medical Genetics</i> , 2010 , 11, 47	2.1	35
133	Wilson Disease in China. <i>Neuroscience Bulletin</i> , 2017 , 33, 323-330	4.3	32
132	The investigation of genetic and clinical features in Chinese patients with juvenile amyotrophic lateral sclerosis. <i>Clinical Genetics</i> , 2017 , 92, 267-273	4	32

131	LRCH1 interferes with DOCK8-Cdc42-induced T cell migration and ameliorates experimental autoimmune encephalomyelitis. <i>Journal of Experimental Medicine</i> , 2017 , 214, 209-226	16.6	29
130	Molecular diagnosis and prophylactic therapy for presymptomatic Chinese patients with Wilson disease. <i>Archives of Neurology</i> , 2003 , 60, 737-41		29
129	Clinical features and mutation spectrum in Chinese patients with CADASIL: A multicenter retrospective study. <i>CNS Neuroscience and Therapeutics</i> , 2017 , 23, 707-716	6.8	25
128	Mutation screening in Chinese patients with familial Alzheimer's disease by whole-exome sequencing. <i>Neurobiology of Aging</i> , 2019 , 76, 215.e15-215.e21	5.6	22
127	Huntington's Disease: Relationship Between Phenotype and Genotype. <i>Molecular Neurobiology</i> , 2017 , 54, 342-348	6.2	21
126	Genetic Spectrum and Variability in Chinese Patients with Amyotrophic Lateral Sclerosis 2019 , 10, 1199-1206		21
125	Identification and functional characterization of two missense mutations in NDRG1 associated with Charcot-Marie-Tooth disease type 4D. <i>Human Mutation</i> , 2017 , 38, 1569-1578	4.7	21
124	Population genetics and new insight into range of CAG repeats of spinocerebellar ataxia type 3 in the Han Chinese population. <i>PLoS ONE</i> , 2015 , 10, e0134405	3.7	21
123	Modeling Neurological Disease by Rapid Conversion of Human Urine Cells into Functional Neurons. <i>Stem Cells International</i> , 2016 , 2016, 2452985	5	20
122	Clinical and genetic spectra in a series of Chinese patients with Charcot-Marie-Tooth disease. <i>Clinica Chimica Acta</i> , 2015 , 451, 263-70	6.2	19
121	Targeted next-generation sequencing improves diagnosis of hereditary spastic paraplegia in Chinese patients. <i>Journal of Molecular Medicine</i> , 2018 , 96, 701-712	5.5	19
120	The discrepancy between the absence of copper deposition and the presence of neuronal damage in the brain of <i>Atp7b</i> (^{-/-}) mice. <i>Metallomics</i> , 2015 , 7, 283-8	4.5	18
119	Sensory nerve disturbance in amyotrophic lateral sclerosis. <i>Life Sciences</i> , 2018 , 203, 242-245	6.8	18
118	Chinese patients with Huntington's disease initially presenting with spinocerebellar ataxia. <i>Clinical Genetics</i> , 2013 , 83, 380-3	4	18
117	Urine-derived induced pluripotent stem cells as a modeling tool for paroxysmal kinesigenic dyskinesia. <i>Biology Open</i> , 2015 , 4, 1744-52	2.2	18
116	Variants of CYP27B1 are associated with both multiple sclerosis and neuromyelitis optica patients in Han Chinese population. <i>Gene</i> , 2015 , 557, 236-9	3.8	18
115	Molecular analyses of GCH-1, TH and parkin genes in Chinese dopa-responsive dystonia families. <i>Clinical Genetics</i> , 2008 , 74, 513-21	4	17
114	Clinical features and outcome in patients with osseomuscular type of Wilson's disease. <i>BMC Neurology</i> , 2017 , 17, 34	3.1	16

113	Associations between apolipoprotein E genotypes and serum levels of glucose, cholesterol, and triglycerides in a cognitively normal aging Han Chinese population. <i>Clinical Interventions in Aging</i> , 2014 , 9, 1063-7	4	16
112	Genotype-phenotype correlation in Chinese patients with spinal and bulbar muscular atrophy. <i>PLoS ONE</i> , 2015 , 10, e0122279	3.7	16
111	The role of CD2AP in the Pathogenesis of Alzheimer's Disease 2019 , 10, 901-907		16
110	Decreased gene expression of CD2AP in Chinese patients with sporadic Alzheimer's disease. <i>Neurobiology of Aging</i> , 2017 , 56, 212.e5-212.e10	5.6	15
109	Identification of 46 CAG repeats within PPP2R2B as probably the shortest pathogenic allele for SCA12. <i>Parkinsonism and Related Disorders</i> , 2015 , 21, 398-401	3.6	15
108	Mutation Analysis of MR-1, SLC2A1, and CLCN1 in 28 PRRT2-negative Paroxysmal Kinesigenic Dyskinesia Patients. <i>Chinese Medical Journal</i> , 2016 , 129, 1017-21	2.9	15
107	Novel Function of Extracellular Matrix Protein 1 in Suppressing Th17 Cell Development in Experimental Autoimmune Encephalomyelitis. <i>Journal of Immunology</i> , 2016 , 197, 1054-64	5.3	15
106	Common GSAP promoter variant contributes to Alzheimer's disease liability. <i>Neurobiology of Aging</i> , 2014 , 35, 2656.e1-2656.e7	5.6	15
105	PRRT2 c.649dupC mutation derived from de novo in paroxysmal kinesigenic dyskinesia. <i>CNS Neuroscience and Therapeutics</i> , 2013 , 19, 61-5	6.8	15
104	FSHD in Chinese population: characteristics of translocation and genotype-phenotype correlation. <i>Neurology</i> , 2004 , 63, 581-3	6.5	15
103	Clinical features and genetic spectrum in Chinese patients with recessive hereditary spastic paraplegia. <i>Translational Neurodegeneration</i> , 2019 , 8, 19	10.3	14
102	Huntington Disease in Asia. <i>Chinese Medical Journal</i> , 2015 , 128, 1815-9	2.9	14
101	Cell type-differential modulation of prefrontal cortical GABAergic interneurons on low gamma rhythm and social interaction. <i>Science Advances</i> , 2020 , 6, eaay4073	14.3	14
100	Genetic spectrum and clinical profiles in a southeast Chinese cohort of Charcot-Marie-Tooth disease. <i>Clinical Genetics</i> , 2019 , 96, 439-448	4	13
99	The polymorphism of the ATP-binding cassette transporter 1 gene modulates Alzheimer disease risk in Chinese Han ethnic population. <i>American Journal of Geriatric Psychiatry</i> , 2012 , 20, 603-11	6.5	13
98	Chinese patients with Machado-Joseph disease presenting with complicated hereditary spastic paraplegia. <i>European Journal of Neurology</i> , 2009 , 16, 953-6	6	13
97	Mitochondrial NADH Dehydrogenase Subunit 3 Polymorphism Associated with an Earlier Age at Onset in Male Machado-Joseph disease Patients. <i>CNS Neuroscience and Therapeutics</i> , 2016 , 22, 38-42	6.8	13
96	Intermediate-length polyglutamine in ATXN2 is a possible risk factor among Eastern Chinese patients with amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2015 , 36, 1603.e11-4	5.6	12

95	Clusterin variants are not associated with southern Chinese patients with Alzheimer's disease. <i>Neurobiology of Aging</i> , 2014 , 35, 2656.e9-2656.e11	5.6	11
94	Mutation Analysis of COQ2 in Chinese Patients with Cerebellar Subtype of Multiple System Atrophy. <i>CNS Neuroscience and Therapeutics</i> , 2015 , 21, 626-30	6.8	11
93	Contribution of intragenic deletions to mutation spectrum in Chinese patients with Wilson's disease and possible mechanism underlying ATP7B gross deletions. <i>Parkinsonism and Related Disorders</i> , 2019 , 62, 128-133	3.6	10
92	Novel PLA2G6 mutations and clinical heterogeneity in Chinese cases with phospholipase A2-associated neurodegeneration. <i>Parkinsonism and Related Disorders</i> , 2018 , 49, 88-94	3.6	10
91	Expert Consensus on Clinical Diagnostic Criteria for Fatal Familial Insomnia. <i>Chinese Medical Journal</i> , 2018 , 131, 1613-1617	2.9	10
90	ABCD1 mutations and phenotype distribution in Chinese patients with X-linked adrenoleukodystrophy. <i>Gene</i> , 2013 , 522, 117-20	3.8	10
89	A Novel Missense Mutation in Peripheral Myelin Protein-22 Causes Charcot-Marie-Tooth Disease. <i>Chinese Medical Journal</i> , 2017 , 130, 1779-1784	2.9	10
88	Variants of Interleukin-7/Interleukin-7 Receptor Alpha are Associated with Both Neuromyelitis Optica and Multiple Sclerosis Among Chinese Han Population in Southeastern China. <i>Chinese Medical Journal</i> , 2015 , 128, 3062-8	2.9	10
87	CCG polymorphisms in the huntingtin gene have no effect on the pathogenesis of patients with Huntington's disease in mainland Chinese families. <i>Journal of the Neurological Sciences</i> , 2012 , 312, 92-6	3.2	10
86	Improving molecular diagnosis of Chinese patients with Charcot-Marie-Tooth by targeted next-generation sequencing and functional analysis. <i>Oncotarget</i> , 2016 , 7, 27655-64	3.3	10
85	Associations between neuroanatomical abnormality and motor symptoms in paroxysmal kinesigenic dyskinesia. <i>Parkinsonism and Related Disorders</i> , 2019 , 62, 134-140	3.6	10
84	Defective roles of ATP7B missense mutations in cellular copper tolerance and copper excretion. <i>Molecular and Cellular Neurosciences</i> , 2015 , 67, 31-6	4.8	9
83	Clinical features of Chinese patients with Huntington's disease carrying CAG repeats beyond 60 within HTT gene. <i>Clinical Genetics</i> , 2014 , 85, 189-93	4	9
82	The role of apolipoprotein E as a risk factor for an earlier age at onset for Machado-Joseph disease is doubtful. <i>PLoS ONE</i> , 2014 , 9, e111356	3.7	9
81	Development of Research on Huntington Disease in China. <i>Neuroscience Bulletin</i> , 2017 , 33, 312-316	4.3	8
80	No association between identified multiple sclerosis non-MHC risk loci and neuromyelitis optica. <i>Neuroscience Bulletin</i> , 2014 , 30, 1036-1044	4.3	8
79	Clinical features of Chinese patients with Gerstmann-Strüssler-Scheinker identified by targeted next-generation sequencing. <i>Neurobiology of Aging</i> , 2017 , 49, 216.e1-216.e5	5.6	8
78	FTL mutation in a Chinese pedigree with neuroferritinopathy. <i>Neurology: Genetics</i> , 2016 , 2, e74	3.8	8

77	Clinical and genetic characteristics of Chinese patients with cerebrotendinous xanthomatosis. <i>Orphanet Journal of Rare Diseases</i> , 2019 , 14, 282	4.2	8
76	Biallelic SORD pathogenic variants cause Chinese patients with distal hereditary motor neuropathy. <i>Npj Genomic Medicine</i> , 2021 , 6, 1	6.2	8
75	A novel WARS mutation causes distal hereditary motor neuropathy in a Chinese family. <i>Brain</i> , 2019 , 142, e49	11.2	7
74	Incidence of Multiple Sclerosis and Related Disorders in Asian Populations of British Columbia. <i>Canadian Journal of Neurological Sciences</i> , 2015 , 42, 235-41	1	7
73	Early Application of Auxiliary Partial Orthotopic Liver Transplantation in Murine Model of Wilson Disease. <i>Transplantation</i> , 2015 , 99, 2317-24	1.8	7
72	Chinese patients with spinocerebellar ataxia type 3 presenting with rare clinical symptoms. <i>Journal of the Neurological Sciences</i> , 2013 , 324, 167-71	3.2	7
71	The discriminative capacity of CSF A β 42 and Tau in neurodegenerative diseases in the Chinese population. <i>Journal of the Neurological Sciences</i> , 2020 , 412, 116756	3.2	6
70	Genetic and clinical features of Chinese patients with mitochondrial ataxia identified by targeted next-generation sequencing. <i>CNS Neuroscience and Therapeutics</i> , 2019 , 25, 21-29	6.8	6
69	Tuberous Sclerosis Complex in Chinese patients: Phenotypic analysis and mutational screening of TSC1/TSC2 genes. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2019 , 71, 322-327	3.2	6
68	EGFP tags affect cellular localization of ATP7B mutants. <i>CNS Neuroscience and Therapeutics</i> , 2013 , 19, 346-51	6.8	6
67	Paroxysmal kinesigenic dyskinesia and myotonia congenita in the same family: coexistence of a PRRT2 mutation and two CLCN1 mutations. <i>Neuroscience Bulletin</i> , 2014 , 30, 1010-1016	4.3	6
66	Clinical and Genetic Profiles in Chinese Patients with Huntington's Disease: A Ten-year Multicenter Study in China 2019 , 10, 1003-1011		6
65	Variant of Associated with Increasing Risk in Chinese Patients with Relapsing-remitting Multiple Sclerosis. <i>Chinese Medical Journal</i> , 2018 , 131, 643-647	2.9	6
64	TMEM151A variants cause paroxysmal kinesigenic dyskinesia. <i>Cell Discovery</i> , 2021 , 7, 83	22.3	6
63	Correlation Between CCG Polymorphisms and CAG Repeats During Germline Transmission in Chinese Patients with Huntington's Disease. <i>Neuroscience Bulletin</i> , 2020 , 36, 811-814	4.3	5
62	Zinc monotherapy and a low-copper diet are beneficial in patients with Wilson disease after liver transplantation. <i>CNS Neuroscience and Therapeutics</i> , 2013 , 19, 905-7	6.8	5
61	A Chinese pedigree with an individual homozygous for CAG repeats of Huntington's disease. <i>Psychiatric Genetics</i> , 2012 , 22, 53-4	2.9	5
60	Gab1 mediates PDGF signaling and is essential to oligodendrocyte differentiation and CNS myelination. <i>ELife</i> , 2020 , 9,	8.9	5

59	Functional study and pathogenicity classification of PRRT2 missense variants in PRRT2-related disorders. <i>CNS Neuroscience and Therapeutics</i> , 2020 , 26, 39-46	6.8	5
58	Genetic spectrum of MCM3AP and its relationship with phenotype of Charcot-Marie-Tooth disease. <i>Journal of the Peripheral Nervous System</i> , 2020 , 25, 107-111	4.7	5
57	Cerebellar spreading depolarization mediates paroxysmal movement disorder. <i>Cell Reports</i> , 2021 , 36, 109743	10.6	5
56	Clinical features and genetic characteristics of homozygous spinocerebellar ataxia type 3. <i>Molecular Genetics & Genomic Medicine</i> , 2020 , 8, e1314	2.3	4
55	Novel GDAP2 pathogenic variants cause autosomal recessive spinocerebellar ataxia-27 (SCAR27) in a Chinese family. <i>Brain</i> , 2020 , 143, e50	11.2	4
54	Phenotypic variance in monozygotic twins with SCA3. <i>Molecular Genetics & Genomic Medicine</i> , 2020 , 8, e1438	2.3	4
53	A novel frameshift ACTN2 variant causes a rare adult-onset distal myopathy with multi-minicores. <i>CNS Neuroscience and Therapeutics</i> , 2021 , 27, 1198-1205	6.8	4
52	Role for Biochemical Assays and Kayser-Fleischer Rings in Diagnosis of Wilson's Disease. <i>Clinical Gastroenterology and Hepatology</i> , 2021 , 19, 590-596	6.9	4
51	Effect of Apolipoprotein E Genotypes on Huntington's Disease Phenotypes in a Han Chinese Population. <i>Neuroscience Bulletin</i> , 2019 , 35, 756-762	4.3	3
50	Genetic Analysis of Chinese Patients with Early-Onset Dementia Using Next-Generation Sequencing. <i>Clinical Interventions in Aging</i> , 2020 , 15, 1831-1839	4	3
49	Primary age-related tauopathy in a Chinese cohort. <i>Journal of Zhejiang University: Science B</i> , 2020 , 21, 256-262	4.5	3
48	Identification and functional characterization of mutations within HADHB associated with mitochondrial trifunctional protein deficiency. <i>Mitochondrion</i> , 2019 , 49, 200-205	4.9	3
47	Lack of association between CALHM1 p.P86L variation and Alzheimer's disease in the Han Chinese population. <i>Neurobiology of Aging</i> , 2014 , 35, 1956.e13-4	5.6	3
46	Novel ATP13A2 and PINK1 variants identified in Chinese patients with Parkinson's disease by whole-exome sequencing. <i>Neuroscience Letters</i> , 2020 , 733, 135075	3.3	3
45	New clinical characteristics and novel pathogenic variants of patients with hereditary leukodystrophies. <i>CNS Neuroscience and Therapeutics</i> , 2020 , 26, 567-575	6.8	3
44	Bi-allelic loss of function variants in COX20 gene cause autosomal recessive sensory neuronopathy. <i>Brain</i> , 2021 , 144, 2457-2470	11.2	3
43	Haplotype analysis encompassing HTT gene in Chinese patients with Huntington's disease. <i>European Journal of Neurology</i> , 2020 , 27, 273-279	6	3
42	7T MRI with post-processing for the presurgical evaluation of pharmacoresistant focal epilepsy. <i>Therapeutic Advances in Neurological Disorders</i> , 2021 , 14, 17562864211021181	6.6	3

41	A novel ceruloplasmin mutation identified in a Chinese patient and clinical spectrum of aceruloplasminemia patients. <i>Metabolic Brain Disease</i> , 2021 , 36, 2273-2281	3.9	3
40	Identification and functional analysis of novel mutations in the gene in Chinese patients with amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2019 , 20, 222-228	3.6	2
39	Characteristic of gut microbiota in southeastern Chinese patients with neuromyelitis optica spectrum disorders. <i>Multiple Sclerosis and Related Disorders</i> , 2020 , 44, 102217	4	2
38	Variants of Interferon Regulatory Factor 5 are Associated with Neither Neuromyelitis Optica Nor Multiple Sclerosis in the Southeastern Han Chinese Population. <i>Chinese Medical Journal</i> , 2015 , 128, 1743-1749	2.9	2
37	Features Differ Between Paroxysmal Kinesigenic Dyskinesia Patients with PRRT2 and TMEM151A Variants.. <i>Movement Disorders</i> , 2022 ,	7	2
36	Genetic association of CUGBP2 and DNMBP with Alzheimer's disease in the Chinese Han population. <i>Current Alzheimer Research</i> , 2015 , 12, 228-32	3	2
35	Genetic spectrum and clinical features in a cohort of Chinese patients with autosomal recessive cerebellar ataxias. <i>Translational Neurodegeneration</i> , 2021 , 10, 40	10.3	2
34	Diffuse intracranial calcification, deep medullary vein engorgement, and symmetric white matter involvement in a patient with systemic lupus erythematosus. <i>CNS Neuroscience and Therapeutics</i> , 2020 , 26, 278-280	6.8	2
33	Genotype-phenotype correlation in 667 Chinese families with spinocerebellar ataxia type 3. <i>Parkinsonism and Related Disorders</i> , 2020 , 78, 116-121	3.6	2
32	Genetic profile and clinical characteristics of Chinese patients with spinocerebellar ataxia type 2: A multicenter experience over 10 years. <i>European Journal of Neurology</i> , 2021 , 28, 955-964	6	2
31	A novel de novo SPTAN1 nonsense variant causes hereditary motor neuropathy in a Chinese family. <i>Brain</i> , 2021 , 144, e11	11.2	2
30	Common genetic variants in PRRC2A are associated with both neuromyelitis optica spectrum disorder and multiple sclerosis in Han Chinese population. <i>Journal of Neurology</i> , 2021 , 268, 506-515	5.5	2
29	Application of Cerebrospinal Fluid AT(N) Framework on the Diagnosis of AD and Related Cognitive Disorders in Chinese Han Population. <i>Clinical Interventions in Aging</i> , 2021 , 16, 311-323	4	2
28	Optimal Combinations of AT(N) Biomarkers to Determine Longitudinal Cognition in the Alzheimer's Disease. <i>Frontiers in Aging Neuroscience</i> , 2021 , 13, 718959	5.3	2
27	Clinical features and outcome of Wilson's disease with generalized epilepsy in Chinese patients. <i>CNS Neuroscience and Therapeutics</i> , 2020 , 26, 842-850	6.8	1
26	A cephalometric study in patients with Wilson's disease. <i>Journal of Clinical Neuroscience</i> , 2019 , 67, 105-108	10.8	1
25	Penetrance estimation of PRRT2 variants in paroxysmal kinesigenic dyskinesia and infantile convulsions. <i>Frontiers of Medicine</i> , 2021 , 1	12	1
24	Three-Dimensional Heterogeneity of Cerebellar Interposed Nucleus-Recipient Zones in the Thalamic Nuclei. <i>Neuroscience Bulletin</i> , 2021 , 37, 1529-1541	4.3	1

23	Factors Associated with Intergenerational Instability of ATXN3 CAG Repeat and Genetic Anticipation in Chinese Patients with Spinocerebellar Ataxia Type 3. <i>Cerebellum</i> , 2020 , 19, 902-906	4.3	1
22	Eomesodermin in CD4T cells is essential for Ginkgolide K ameliorating disease progression in experimental autoimmune encephalomyelitis. <i>International Journal of Biological Sciences</i> , 2021 , 17, 50-61 ^{11.2}	11.2	1
21	CHIP control degradation of mutant ETF:QO through ubiquitylation in late-onset multiple acyl-CoA dehydrogenase deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2021 , 44, 450-468	5.4	1
20	Prevalent Pathogenic Variants of in Chinese Patients with Wilson's Disease: Geographical Distribution and Founder Effect. <i>Genes</i> , 2021 , 12,	4.2	1
19	A novel PCYT2 mutation identified in a Chinese consanguineous family with hereditary spastic paraplegia. <i>Journal of Genetics and Genomics</i> , 2021 , 48, 751-754	4	1
18	Novel bi-allelic HPDL variants cause hereditary spastic paraplegia in a Chinese patient. <i>Clinical Genetics</i> , 2021 , 100, 777-778	4	1
17	The clinical, imaging and biological features of psychosis in Han Chinese patients with Huntington's disease. <i>Journal of Psychiatric Research</i> , 2021 , 141, 333-338	5.2	1
16	Paroxysmal Kinesigenic Dyskinesia Caused by 16p11.2 Microdeletion and Related Clinical Features.. <i>Neurology: Genetics</i> , 2022 , 8, e659	3.8	1
15	Identification and functional characterization of novel GDAP1 variants in Chinese patients with Charcot-Marie-Tooth disease. <i>Annals of Clinical and Translational Neurology</i> , 2020 , 7, 2381-2392	5.3	0
14	Taste loss as the sole presenting symptom in Chinese patient with facial onset sensory and motor neuropathy. <i>CNS Neuroscience and Therapeutics</i> , 2021 , 27, 1610-1613	6.8	0
13	A de novo variant of POLR3B causes demyelinating Charcot-Marie-Tooth disease in a Chinese patient: a case report. <i>BMC Neurology</i> , 2021 , 21, 402	3.1	0
12	Mutation Spectrum and Natural History of ALS Patients in a 15-Year Cohort in Southeastern China. <i>Frontiers in Genetics</i> , 2021 , 12, 746060	4.5	0
11	Identification of pathogenic C9orf72 hexanucleotide repeat expansion in a Chinese patient with frontotemporal dementia: A case report. <i>CNS Neuroscience and Therapeutics</i> , 2021 , 27, 725-727	6.8	0
10	TGM6 might not be a specific causative gene for spinocerebellar ataxia resulting from genetic analysis and functional study. <i>Gene</i> , 2021 , 779, 145495	3.8	0
9	Spastic paraplegia as the only symptom in two adult-onset patients carrying a novel pathogenic variant in PYCR2. <i>European Journal of Neurology</i> , 2021 , 28, e17-e19	6	0
8	Clinical Characterization and Founder Effect Analysis in Chinese Patients with Phospholipase A2-Associated Neurodegeneration. <i>Brain Sciences</i> , 2022 , 12, 517	3.4	0
7	Identification of susceptibility loci for cognitive impairment in a cohort of Han Chinese patients with Parkinson's disease. <i>Neuroscience Letters</i> , 2020 , 730, 135034	3.3	
6	Alzheimer's disease susceptibility locus in CD2AP is associated with increased cerebrospinal fluid tau levels in mild cognitive impairment.. <i>Neuroscience Letters</i> , 2021 , 136419	3.3	

- 5 A Mutation in Dystrophin Causing Muscular Dystrophy in a Female Patient. *Chinese Medical Journal*, **2017**, 130, 2273-2278 2.9
- 4 Emerging neurological symptoms after liver transplantation: A 6-year follow-up of an adolescent patient with Wilson's disease.. *CNS Neuroscience and Therapeutics*, **2022**, 6.8
- 3 Clinical and genetic characteristics of Chinese patients with reducing body myopathy. *Neuromuscular Disorders*, **2021**, 31, 442-449 2.9
- 2 Identification of a large homozygous SPG21 deletion in a Chinese patient with Mast syndrome. *CNS Neuroscience and Therapeutics*, **2021**, 27, 1251-1253 6.8
- 1 A novel UBAP1 truncated variant in a Chinese family with hereditary spastic paraplegia.. *Molecular Genetics & Genomic Medicine*, **2022**, e1927 2.3