Zhi-Ying Wu

List of Publications by Year in Descending Order

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	Features Differ Between Paroxysmal Kinesigenic Dyskinesia Patients with PRRT2 and TMEM151A Variants.. *Movement Disorders*, **2022**,	7	2
147	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	
146	Paroxysmal Kinesigenic Dyskinesia Caused by 16p11.2 Microdeletion and Related Clinical Features.. *Neurology: Genetics*, **2022**, 8, e659	3.8	1
145	A novel UBAP1 truncated variant in a Chinese family with hereditary spastic paraplegia.. *Molecular Genetics & Genomic Medicine*, **2022**, e1927	2.3	
144	Clinical Characterization and Founder Effect Analysis in Chinese Patients with Phospholipase A2-Associated Neurodegeneration. *Brain Sciences*, **2022**, 12, 517	3.4	0
143	Alzheimer's disease susceptibility locus in CD2AP is associated with increased cerebrospinal fluid tau levels in mild cognitive impairment.. *Neuroscience Letters*, **2021**, 136419	3.3	
142	Taste loss as the sole presenting symptom in Chinese patient with facial onset sensory and motor neuronopathy. *CNS Neuroscience and Therapeutics*, **2021**, 27, 1610-1613	6.8	0
141	Penetrance estimation of PRRT2 variants in paroxysmal kinesigenic dyskinesia and infantile convulsions. *Frontiers of Medicine*, **2021**, 1	12	1
140	Genetic spectrum and clinical features in a cohort of Chinese patients with autosomal recessive cerebellar ataxias. *Translational Neurodegeneration*, **2021**, 10, 40	10.3	2
139	A de novo variant of POLR3B causes demyelinating Charcot-Marie-Tooth disease in a Chinese patient: a case report. *BMC Neurology*, **2021**, 21, 402	3.1	0
138	Three-Dimensional Heterogeneity of Cerebellar Interposed Nucleus-Recipient Zones in the Thalamic Nuclei. *Neuroscience Bulletin*, **2021**, 37, 1529-1541	4.3	1
137	Mutation Spectrum and Natural History of ALS Patients in a 15-Year Cohort in Southeastern China. *Frontiers in Genetics*, **2021**, 12, 746060	4.5	0
136	Bi-allelic loss of function variants in COX20 gene cause autosomal recessive sensory neuronopathy. *Brain*, **2021**, 144, 2457-2470	11.2	3
135	Identification of pathogenic C9orf72 hexanucleotide repeat expansion in a Chinese patient with frontotemporal dementia: A case report. *CNS Neuroscience and Therapeutics*, **2021**, 27, 725-727	6.8	0
134	TGM6 might not be a specific causative gene for spinocerebellar ataxia resulting from genetic analysis and functional study. *Gene*, **2021**, 779, 145495	3.8	0
133	A novel frameshift ACTN2 variant causes a rare adult-onset distal myopathy with multi-minicores. *CNS Neuroscience and Therapeutics*, **2021**, 27, 1198-1205	6.8	4
132	Role for Biochemical Assays and Kayser-Fleischer Rings in Diagnosis of Wilson's Disease. *Clinical Gastroenterology and Hepatology*, **2021**, 19, 590-596	6.9	4

131	Genetic profile and clinical characteristics of Chinese patients with spinocerebellar ataxia type 2: A multicenter experience over 10 years. *European Journal of Neurology*, **2021**, 28, 955-964	6	2
130	A novel de novo SPTAN1 nonsense variant causes hereditary motor neuropathy in a Chinese family. *Brain*, **2021**, 144, e11	11.2	2
129	Eomesodermin in CD4T cells is essential for Ginkgolide K ameliorating disease progression in experimental autoimmune encephalomyelitis. *International Journal of Biological Sciences*, **2021**, 17, 50-61	11.2	1
128	Spastic paraplegia as the only symptom in two adult-onset patients carrying a novel pathogenic variant in PYCR2. *European Journal of Neurology*, **2021**, 28, e17-e19	6	0
127	Common genetic variants in PRRC2A are associated with both neuromyelitis optica spectrum disorder and multiple sclerosis in Han Chinese population. *Journal of Neurology*, **2021**, 268, 506-515	5.5	2
126	CHIP control degradation of mutant ETF:QO through ubiquitylation in late-onset multiple acyl-CoA dehydrogenase deficiency. *Journal of Inherited Metabolic Disease*, **2021**, 44, 450-468	5.4	1
125	7T MRI with post-processing for the presurgical evaluation of pharmacoresistant focal epilepsy. *Therapeutic Advances in Neurological Disorders*, **2021**, 14, 17562864211021181	6.6	3
124	Biallelic SORD pathogenic variants cause Chinese patients with distal hereditary motor neuropathy. *Npj Genomic Medicine*, **2021**, 6, 1	6.2	8
123	Clinical and genetic characteristics of Chinese patients with reducing body myopathy. *Neuromuscular Disorders*, **2021**, 31, 442-449	2.9	
122	Prevalent Pathogenic Variants of in Chinese Patients with Wilson Disease: Geographical Distribution and Founder Effect. *Genes*, **2021**, 12,	4.2	1
121	Application of Cerebrospinal Fluid AT(N) Framework on the Diagnosis of AD and Related Cognitive Disorders in Chinese Han Population. *Clinical Interventions in Aging*, **2021**, 16, 311-323	4	2
120	Optimal Combinations of AT(N) Biomarkers to Determine Longitudinal Cognition in the Alzheimer Disease. *Frontiers in Aging Neuroscience*, **2021**, 13, 718959	5.3	2
119	A novel PCYT2 mutation identified in a Chinese consanguineous family with hereditary spastic paraplegia. *Journal of Genetics and Genomics*, **2021**, 48, 751-754	4	1
118	A novel ceruloplasmin mutation identified in a Chinese patient and clinical spectrum of aceruloplasminemia patients. *Metabolic Brain Disease*, **2021**, 36, 2273-2281	3.9	3
117	TMEM151A variants cause paroxysmal kinesigenic dyskinesia. *Cell Discovery*, **2021**, 7, 83	22.3	6
116	Cerebellar spreading depolarization mediates paroxysmal movement disorder. *Cell Reports*, **2021**, 36, 109743	10.6	5
115	Identification of a large homozygous SPG21 deletion in a Chinese patient with Mast syndrome. *CNS Neuroscience and Therapeutics*, **2021**, 27, 1251-1253	6.8	
114	Novel bi-allelic HPDL variants cause hereditary spastic paraplegia in a Chinese patient. *Clinical Genetics*, **2021**, 100, 777-778	4	1

113	The clinical, imaging and biological features of psychosis in Han Chinese patients with Huntingtonℝ disease. *Journal of Psychiatric Research*, **2021**, 141, 333-338	5.2	1
112	Genetic Analysis of Chinese Patients with Early-Onset Dementia Using Next-Generation Sequencing. *Clinical Interventions in Aging*, **2020**, 15, 1831-1839	4	3
111	Identification and functional characterization of novel GDAP1 variants in Chinese patients with Charcot-Marie-Tooth disease. *Annals of Clinical and Translational Neurology*, **2020**, 7, 2381-2392	5.3	0
110	Identification of susceptibility loci for cognitive impairment in a cohort of Han Chinese patients with Parkinsonℝ disease. *Neuroscience Letters*, **2020**, 730, 135034	3.3	
109	Characteristic of gut microbiota in southeastern Chinese patients with neuromyelitis optica spectrum disorders. *Multiple Sclerosis and Related Disorders*, **2020**, 44, 102217	4	2
108	Primary age-related tauopathy in a Chinese cohort. *Journal of Zhejiang University: Science B*, **2020**, 21, 256-262	4.5	3
107	Correlation Between CCG Polymorphisms and CAG Repeats During Germline Transmission in Chinese Patients with Huntingtonℝ Disease. *Neuroscience Bulletin*, **2020**, 36, 811-814	4.3	5
106	Clinical features and genetic characteristics of homozygous spinocerebellar ataxia type 3. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1314	2.3	4
105	The discriminative capacity of CSF □amyloid 42 and Tau in neurodegenerative diseases in the Chinese population. *Journal of the Neurological Sciences*, **2020**, 412, 116756	3.2	6
104	Clinical features and outcome of Wilsonℝ disease with generalized epilepsy in Chinese patients. *CNS Neuroscience and Therapeutics*, **2020**, 26, 842-850	6.8	1
103	Gab1 mediates PDGF signaling and is essential to oligodendrocyte differentiation and CNS myelination. *ELife*, **2020**, 9,	8.9	5
102	Novel GDAP2 pathogenic variants cause autosomal recessive spinocerebellar ataxia-27 (SCAR27) in a Chinese family. *Brain*, **2020**, 143, e50	11.2	4
101	Novel ATP13A2 and PINK1 variants identified in Chinese patients with Parkinsonℝ disease by whole-exome sequencing. *Neuroscience Letters*, **2020**, 733, 135075	3.3	3
100	Diffuse intracranial calcification, deep medullary vein engorgement, and symmetric white matter involvement in a patient with systemic lupus erythematosus. *CNS Neuroscience and Therapeutics*, **2020**, 26, 278-280	6.8	2
99	New clinical characteristics and novel pathogenic variants of patients with hereditary leukodystrophies. *CNS Neuroscience and Therapeutics*, **2020**, 26, 567-575	6.8	3
98	Genotype-phenotype correlation in 667 Chinese families with spinocerebellar ataxia type 3. *Parkinsonism and Related Disorders*, **2020**, 78, 116-121	3.6	2
97	Factors Associated with Intergenerational Instability of ATXN3 CAG Repeat and Genetic Anticipation in Chinese Patients with Spinocerebellar Ataxia Type 3. *Cerebellum*, **2020**, 19, 902-906	4.3	1
96	Cell type-differential modulation of prefrontal cortical GABAergic interneurons on low gamma rhythm and social interaction. *Science Advances*, **2020**, 6, eaay4073	14.3	14

95	Phenotypic variance in monozygotic twins with SCA3. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1438	2.3	4
94	Functional study and pathogenicity classification of PRRT2 missense variants in PRRT2-related disorders. *CNS Neuroscience and Therapeutics*, **2020**, 26, 39-46	6.8	5
93	Haplotype analysis encompassing HTT gene in Chinese patients with Huntington's disease. *European Journal of Neurology*, **2020**, 27, 273-279	6	3
92	Genetic spectrum of MCM3AP and its relationship with phenotype of Charcot-Marie-Tooth disease. *Journal of the Peripheral Nervous System*, **2020**, 25, 107-111	4.7	5
91	Contribution of intragenic deletions to mutation spectrum in Chinese patients with Wilson's disease and possible mechanism underlying ATP7B gross deletions. *Parkinsonism and Related Disorders*, **2019**, 62, 128-133	3.6	10
90	Identification and functional analysis of novel mutations in the gene in Chinese patients with amyotrophic lateral sclerosis. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2019**, 20, 222-228	3.6	2
89	Effect of Apolipoprotein E Genotypes on Huntington's Disease Phenotypes in a Han Chinese Population. *Neuroscience Bulletin*, **2019**, 35, 756-762	4.3	3
88	Genetic and clinical features of Chinese patients with mitochondrial ataxia identified by targeted next-generation sequencing. *CNS Neuroscience and Therapeutics*, **2019**, 25, 21-29	6.8	6
87	Genetic spectrum and clinical profiles in a southeast Chinese cohort of Charcot-Marie-Tooth disease. *Clinical Genetics*, **2019**, 96, 439-448	4	13
86	Genetic Spectrum and Variability in Chinese Patients with Amyotrophic Lateral Sclerosis **2019**, 10, 1199-1206		21
85	A novel WARS mutation causes distal hereditary motor neuropathy in a Chinese family. *Brain*, **2019**, 142, e49	11.2	7
84	A cephalometric study in patients with Wilson's disease. *Journal of Clinical Neuroscience*, **2019**, 67, 105-108	2.2	1
83	Clinical features and genetic spectrum in Chinese patients with recessive hereditary spastic paraplegia. *Translational Neurodegeneration*, **2019**, 8, 19	10.3	14
82	Tuberous Sclerosis Complex in Chinese patients: Phenotypic analysis and mutational screening of TSC1/TSC2 genes. *Seizure: the Journal of the British Epilepsy Association*, **2019**, 71, 322-327	3.2	6
81	Neurofilament light chain is a promising serum biomarker in spinocerebellar ataxia type 3. *Molecular Neurodegeneration*, **2019**, 14, 39	19	35
80	Identification and functional characterization of mutations within HADHB associated with mitochondrial trifunctional protein deficiency. *Mitochondrion*, **2019**, 49, 200-205	4.9	3
79	Clinical and Genetic Profiles in Chinese Patients with Huntington's Disease: A Ten-year Multicenter Study in China **2019**, 10, 1003-1011		6
78	The role of CD2AP in the Pathogenesis of Alzheimer's Disease **2019**, 10, 901-907		16

77	Clinical and genetic characteristics of Chinese patients with cerebrotendinous xanthomatosis. *Orphanet Journal of Rare Diseases*, **2019**, 14, 282	4.2	8
76	Mutation screening in Chinese patients with familial Alzheimer's disease by whole-exome sequencing. *Neurobiology of Aging*, **2019**, 76, 215.e15-215.e21	5.6	22
75	Associations between neuroanatomical abnormality and motor symptoms in paroxysmal kinesigenic dyskinesia. *Parkinsonism and Related Disorders*, **2019**, 62, 134-140	3.6	10
74	Novel PLA2G6 mutations and clinical heterogeneity in Chinese cases with phospholipase A2-associated neurodegeneration. *Parkinsonism and Related Disorders*, **2018**, 49, 88-94	3.6	10
73	Sensory nerve disturbance in amyotrophic lateral sclerosis. *Life Sciences*, **2018**, 203, 242-245	6.8	18
72	PRRT2 deficiency induces paroxysmal kinesigenic dyskinesia by regulating synaptic transmission in cerebellum. *Cell Research*, **2018**, 28, 90-110	24.7	48
71	Expert Consensus on Clinical Diagnostic Criteria for Fatal Familial Insomnia. *Chinese Medical Journal* , **2018**, 131, 1613-1617	2.9	10
70	Targeted next-generation sequencing improves diagnosis of hereditary spastic paraplegia in Chinese patients. *Journal of Molecular Medicine*, **2018**, 96, 701-712	5.5	19
69	Variant of Associated with Increasing Risk in Chinese Patients with Relapsing-remitting Multiple Sclerosis. *Chinese Medical Journal*, **2018**, 131, 643-647	2.9	6
68	Huntington's Disease: Relationship Between Phenotype and Genotype. *Molecular Neurobiology*, **2017**, 54, 342-348	6.2	21
67	Development of Research on Huntington Disease in China. *Neuroscience Bulletin*, **2017**, 33, 312-316	4.3	8
66	Wilson's Disease in China. *Neuroscience Bulletin*, **2017**, 33, 323-330	4.3	32
65	Decreased gene expression of CD2AP in Chinese patients with sporadic Alzheimer's disease. *Neurobiology of Aging*, **2017**, 56, 212.e5-212.e10	5.6	15
64	The investigation of genetic and clinical features in Chinese patients with juvenile amyotrophic lateral sclerosis. *Clinical Genetics*, **2017**, 92, 267-273	4	32
63	LRCH1 interferes with DOCK8-Cdc42-induced T cell migration and ameliorates experimental autoimmune encephalomyelitis. *Journal of Experimental Medicine*, **2017**, 214, 209-226	16.6	29
62	Clinical features and mutation spectrum in Chinese patients with CADASIL: A multicenter retrospective study. *CNS Neuroscience and Therapeutics*, **2017**, 23, 707-716	6.8	25
61	Identification and functional characterization of two missense mutations in NDRG1 associated with Charcot-Marie-Tooth disease type 4D. *Human Mutation*, **2017**, 38, 1569-1578	4.7	21
60	Clinical features and outcome in patients with osseomuscular type of Wilson's disease. *BMC Neurology*, **2017**, 17, 34	3.1	16

59	Clinical features of Chinese patients with Gerstmann-Str□ssler-Scheinker identified by targeted next-generation sequencing. *Neurobiology of Aging*, **2017**, 49, 216.e1-216.e5	5.6	8
58	A Novel Missense Mutation in Peripheral Myelin Protein-22 Causes Charcot-Marie-Tooth Disease. *Chinese Medical Journal*, **2017**, 130, 1779-1784	2.9	10
57	A Mutation in Dystrophin Causing Muscular Dystrophy in a Female Patient. *Chinese Medical Journal*, **2017**, 130, 2273-2278	2.9	
56	Mutation Analysis of MR-1, SLC2A1, and CLCN1 in 28 PRRT2-negative Paroxysmal Kinesigenic Dyskinesia Patients. *Chinese Medical Journal*, **2016**, 129, 1017-21	2.9	15
55	Novel Function of Extracellular Matrix Protein 1 in Suppressing Th17 Cell Development in Experimental Autoimmune Encephalomyelitis. *Journal of Immunology*, **2016**, 197, 1054-64	5.3	15
54	Spinocerebellar ataxia: relationship between phenotype and genotype - a review. *Clinical Genetics*, **2016**, 90, 305-14	4	79
53	Improving molecular diagnosis of Chinese patients with Charcot-Marie-Tooth by targeted next-generation sequencing and functional analysis. *Oncotarget*, **2016**, 7, 27655-64	3.3	10
52	Modeling Neurological Disease by Rapid Conversion of Human Urine Cells into Functional Neurons. *Stem Cells International*, **2016**, 2016, 2452985	5	20
51	Spectrum and Classification of ATP7B Variants in a Large Cohort of Chinese Patients with Wilson□s Disease Guides Genetic Diagnosis. *Theranostics*, **2016**, 6, 638-49	12.1	49
50	Mitochondrial NADH Dehydrogenase Subunit 3 Polymorphism Associated with an Earlier Age at Onset in Male Machado-Joseph disease Patients. *CNS Neuroscience and Therapeutics*, **2016**, 22, 38-42	6.8	13
49	Near-Infrared Upconversion Chemodosimeter for In Vivo Detection of Cu(2+) in Wilson Disease. *Advanced Materials*, **2016**, 28, 6625-30	24	89
48	FTL mutation in a Chinese pedigree with neuroferritinopathy. *Neurology: Genetics*, **2016**, 2, e74	3.8	8
47	Genotype-phenotype correlations of amyotrophic lateral sclerosis. *Translational Neurodegeneration* , **2016**, 5, 3	10.3	43
46	Defective roles of ATP7B missense mutations in cellular copper tolerance and copper excretion. *Molecular and Cellular Neurosciences*, **2015**, 67, 31-6	4.8	9
45	Identification of 46 CAG repeats within PPP2R2B as probably the□shortest pathogenic allele for SCA12. *Parkinsonism and Related Disorders*, **2015**, 21, 398-401	3.6	15
44	Intermediate-length polyglutamine in ATXN2 is a possible risk factor among Eastern Chinese patients with amyotrophic lateral sclerosis. *Neurobiology of Aging*, **2015**, 36, 1603.e11-4	5.6	12
43	The discrepancy between the absence of copper deposition and the presence of neuronal damage in the brain of Atp7b(-/-) mice. *Metallomics*, **2015**, 7, 283-8	4.5	18
42	Clinical and genetic spectra in a series of Chinese patients with Charcot-Marie-Tooth disease. *Clinica Chimica Acta*, **2015**, 451, 263-70	6.2	19

41	Incidence of Multiple Sclerosis and Related Disorders in Asian Populations of British Columbia. *Canadian Journal of Neurological Sciences*, **2015**, 42, 235-41	1	7
40	Mutation Analysis of COQ2 in Chinese Patients with Cerebellar Subtype of Multiple System Atrophy. *CNS Neuroscience and Therapeutics*, **2015**, 21, 626-30	6.8	11
39	Early Application of Auxiliary Partial Orthotopic Liver Transplantation in Murine Model of Wilson Disease. *Transplantation*, **2015**, 99, 2317-24	1.8	7
38	Variants of Interferon Regulatory Factor 5 are Associated with Neither Neuromyelitis Optica Nor Multiple Sclerosis in the Southeastern Han Chinese Population. *Chinese Medical Journal*, **2015**, 128, 1743-7	2.9	2
37	Variants of Interleukin-7/Interleukin-7 Receptor Alpha are Associated with Both Neuromyelitis Optica and Multiple Sclerosis Among Chinese Han Population in Southeastern China. *Chinese Medical Journal*, **2015**, 128, 3062-8	2.9	10
36	Population genetics and new insight into range of CAG repeats of spinocerebellar ataxia type 3 in the Han Chinese population. *PLoS ONE*, **2015**, 10, e0134405	3.7	21
35	A striatal-enriched intronic GPCR modulates huntingtin levels and toxicity. *ELife*, **2015**, 4,	8.9	49
34	Huntington Disease in Asia. *Chinese Medical Journal*, **2015**, 128, 1815-9	2.9	14
33	Urine-derived induced pluripotent stem cells as a modeling tool for paroxysmal kinesigenic dyskinesia. *Biology Open*, **2015**, 4, 1744-52	2.2	18
32	Variants of CYP27B1 are associated with both multiple sclerosis and neuromyelitis optica patients in Han Chinese population. *Gene*, **2015**, 557, 236-9	3.8	18
31	Genotype-phenotype correlation in Chinese patients with spinal and bulbar muscular atrophy. *PLoS ONE*, **2015**, 10, e0122279	3.7	16
30	Risk prediction for sporadic Alzheimer's disease using genetic risk score in the Han Chinese population. *Oncotarget*, **2015**, 6, 36955-64	3.3	44
29	Genetic association of CUGBP2 and DNMBP with Alzheimer's disease in the Chinese Han population. *Current Alzheimer Research*, **2015**, 12, 228-32	3	2
28	No association between identified multiple sclerosis non-MHC risk loci and neuromyelitis optica. *Neuroscience Bulletin*, **2014**, 30, 1036-1044	4.3	8
27	Lack of association between CALHM1 p.P86L variation and Alzheimer's disease in the Han Chinese population. *Neurobiology of Aging*, **2014**, 35, 1956.e13-4	5.6	3
26	Common GSAP promoter variant contributes to Alzheimer's disease liability. *Neurobiology of Aging*, **2014**, 35, 2656.e1-2656.e7	5.6	15
25	Clusterin variants are not associated with southern Chinese patients with Alzheimer's disease. *Neurobiology of Aging*, **2014**, 35, 2656.e9-2656.e11	5.6	11
24	Clinical features of Chinese patients with Huntington's disease carrying CAG repeats beyond 60 within HTT gene. *Clinical Genetics*, **2014**, 85, 189-93	4	9

23	The role of apolipoprotein E as a risk factor for an earlier age at onset for Machado-Joseph disease is doubtful. *PLoS ONE*, **2014**, 9, e111356	3.7	9
22	Associations between apolipoprotein E genotypes and serum levels of glucose, cholesterol, and triglycerides in a cognitively normal aging Han Chinese population. *Clinical Interventions in Aging*, **2014**, 9, 1063-7	4	16
21	Paroxysmal kinesigenic dyskinesia and myotonia congenita in the same family: coexistence of a PRRT2 mutation and two CLCN1 mutations. *Neuroscience Bulletin*, **2014**, 30, 1010-1016	4.3	6
20	Identify mutation in amyotrophic lateral sclerosis cases using HaloPlex target enrichment system. *Neurobiology of Aging*, **2014**, 35, 2881.e11-2881.e15	5.6	38
19	EGFP tags affect cellular localization of ATP7B mutants. *CNS Neuroscience and Therapeutics*, **2013**, 19, 346-51	6.8	6
18	ABCD1 mutations and phenotype distribution in Chinese patients with X-linked adrenoleukodystrophy. *Gene*, **2013**, 522, 117-20	3.8	10
17	PRRT2 c.649dupC mutation derived from de novo in paroxysmal kinesigenic dyskinesia. *CNS Neuroscience and Therapeutics*, **2013**, 19, 61-5	6.8	15
16	Chinese patients with Huntington's disease initially presenting with spinocerebellar ataxia. *Clinical Genetics*, **2013**, 83, 380-3	4	18
15	Chinese patients with spinocerebellar ataxia type 3 presenting with rare clinical symptoms. *Journal of the Neurological Sciences*, **2013**, 324, 167-71	3.2	7
14	PRRT2 mutation correlated with phenotype of paroxysmal kinesigenic dyskinesia and drug response. *Neurology*, **2013**, 80, 1534-5	6.5	47
13	Zinc monotherapy and a low-copper diet are beneficial in patients with Wilson disease after liver transplantation. *CNS Neuroscience and Therapeutics*, **2013**, 19, 905-7	6.8	5
12	CCG polymorphisms in the huntingtin gene have no effect on the pathogenesis of patients with Huntington's disease in mainland Chinese families. *Journal of the Neurological Sciences*, **2012**, 312, 92-6	3.2	10
11	The polymorphism of the ATP-binding cassette transporter 1 gene modulates Alzheimer disease risk in Chinese Han ethnic population. *American Journal of Geriatric Psychiatry*, **2012**, 20, 603-11	6.5	13
10	A Chinese pedigree with an individual homozygous for CAG repeats of Huntington's disease. *Psychiatric Genetics*, **2012**, 22, 53-4	2.9	5
9	Exome sequencing identifies truncating mutations in PRRT2 that cause paroxysmal kinesigenic dyskinesia. *Nature Genetics*, **2011**, 43, 1252-5	36.3	339
8	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. *Journal of Molecular Medicine*, **2011**, 89, 569-76	5.5	53
7	High frequency of Machado-Joseph disease identified in southeastern Chinese kindreds with spinocerebellar ataxia. *BMC Medical Genetics*, **2010**, 11, 47	2.1	35
6	Chinese patients with Machado-Joseph disease presenting with complicated hereditary spastic paraplegia. *European Journal of Neurology*, **2009**, 16, 953-6	6	13

5	Molecular analyses of GCH-1, TH and parkin genes in Chinese dopa-responsive dystonia families. *Clinical Genetics*, **2008**, 74, 513-21	4	17
4	Mutation analysis of 218 Chinese patients with Wilson disease revealed no correlation between the canine copper toxicosis gene MURR1 and Wilson disease. *Journal of Molecular Medicine*, **2006**, 84, 438-42	5.5	44
3	FSHD in Chinese population: characteristics of translocation and genotype-phenotype correlation. *Neurology*, **2004**, 63, 581-3	6.5	15
2	Molecular diagnosis and prophylactic therapy for presymptomatic Chinese patients with Wilson disease. *Archives of Neurology*, **2003**, 60, 737-41		29
1	Mutation analysis and the correlation between genotype and phenotype of Arg778Leu mutation in chinese patients with Wilson disease. *Archives of Neurology*, **2001**, 58, 971-6		67