## Ning Wang

## List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Prevalence and disease progression of genetically-confirmed facioscapulohumeral muscular dystrophy type 1 (FSHD1) in China between 2001 and 2020: a nationwide population-based study. The Lancet Regional Health - Western Pacific, 2022, 18, 100323.	1.3	10
2	Cutaneous Lesions as a Clue to the Etiology of Extensive Intracranial Calcifications: Aicardi-Goutià res Syndrome. Neurology, 2022, , 10.1212/WNL.00000000013294.	1.5	0
3	Chinese patients with hereditary spastic paraplegias (HSPs): a protocol for a hospital-based cohort study. BMJ Open, 2022, 12, e054011.	0.8	O
4	Wheelchair use in genetically confirmed FSHD1 from a large cohort study in Chinese population. Brain, 2022, 145, e51-e54.	3.7	3
5	Generation and characterization of an induced pluripotent stem cell line (FJMUNi001-A) from a patient with Duchenne muscular dystrophy carrying c.4518Â+Â512ÂTÂ>ÂA variant in the DMD gene. Stem Cell Research, 2022, 60, 102718.	0.3	O
6	Exomeâ€Wide Analyses in Paroxysmal Kinesigenic Dyskinesia Confirm <scp><i>TMEM151A</i></scp> as a Novel Causative Gene. Movement Disorders, 2022, 37, 641-643.	2.2	10
7	Clinical Characterization and Founder Effect Analysis in Chinese Patients with Phospholipase A2-Associated Neurodegeneration. Brain Sciences, 2022, 12, 517.	1.1	4
8	CRISPR/Cas9-based genome editing for the modification of multiple duplications that cause Duchenne muscular dystrophy. Gene Therapy, 2022, 29, 730-737.	2.3	3
9	Expanding the phenotype and genotype spectra of PLIN4-associated myopathy with rimmed ubiquitin-positive autophagic vacuolation. Acta Neuropathologica, 2022, 143, 733-735.	3.9	6
10	<scp>GGC</scp> Repeat Expansion of <scp><i>RILPL1</i></scp> is Associated with Oculopharyngodistal Myopathy. Annals of Neurology, 2022, 92, 512-526.	2.8	16
11	Genetic variations and clinical spectrum of dystroglycanopathy in a large cohort of Chinese patients. Clinical Genetics, 2021, 99, 384-395.	1.0	19
12	<scp>CHIP</scp> control degradation of mutant <scp>ETF</scp> : <scp>QO</scp> through ubiquitylation in lateâ€onset multiple <scp>acylâ€CoA</scp> dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 450-468.	1.7	3
13	Decreased serum creatinine levels predict short survival in amyotrophic lateral sclerosis. Annals of Clinical and Translational Neurology, 2021, 8, 448-455.	1.7	12
14	Recommendations for the diagnosis and treatment of paroxysmal kinesigenic dyskinesia: an expert consensus in China. Translational Neurodegeneration, 2021, 10, 7.	3.6	19
15	Serum Uric Acid Levels Predict Mortality Risk in Male Amyotrophic Lateral Sclerosis Patients. Frontiers in Neurology, 2021, 12, 602663.	1.1	7
16	A novel start codon variant in SMCHD1 from a Chinese family causes facioscapulohumeral muscular dystrophy type 2. Chinese Medical Journal, 2021, Publish Ahead of Print, 2753-2755.	0.9	0
17	Establishment of a human iPSC line XMDYYYi001-A from a patient with Becker muscular dystrophy harboring duplications of exons 2-19 in dystrophin gene. Stem Cell Research, 2021, 53, 102298.	0.3	O
18	Potential markers for sample size estimations in hereditary spastic paraplegia type 5. Orphanet Journal of Rare Diseases, 2021, 16, 391.	1.2	2

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19	Exhausting T Cells During HIV Infection May Improve the Prognosis of Patients with COVID-19. Frontiers in Cellular and Infection Microbiology, 2021, 11, 564938.	1.8	2
20	Higher Concentration of Plasma <scp>Glial Fibrillary Acidic Protein</scp> in Wilson Disease Patients with Neurological Manifestations. Movement Disorders, 2021, 36, 1446-1450.	2.2	9
21	Genetic screening method for analyzing survival motor neuron copy number in spinal muscular atrophy by multiplex ligation-dependent probe amplification and droplet digital polymerase chain reaction. Chinese Medical Journal, 2020, 133, 2510-2511.	0.9	2
22	Promoting identification of amyotrophic lateral sclerosis based on labelâ€free plasma spectroscopy. Annals of Clinical and Translational Neurology, 2020, 7, 2010-2018.	1.7	9
23	Median Nerve-Neurophysiological Index Correlates With the Survival of Patients With Amyotrophic Lateral Sclerosis. Frontiers in Neurology, 2020, 11, 570227.	1.1	4
24	Novel <i>CAPN1</i> mutations extend the phenotypic heterogeneity in combined spastic paraplegia and ataxia. Annals of Clinical and Translational Neurology, 2020, 7, 1862-1869.	1.7	11
25	Clinical and genetic features of somatic mosaicism in facioscapulohumeral dystrophy. Journal of Medical Genetics, 2020, 57, 777-785.	1.5	7
26	Quantitative assessment of postural instability in spinocerebellar ataxia type 3 patients. Annals of Clinical and Translational Neurology, 2020, 7, 1360-1370.	1.7	7
27	The impact of ethnicity on the clinical presentations of spinocerebellar ataxia type 3. Parkinsonism and Related Disorders, 2020, 72, 37-43.	1.1	16
28	Cryptic exon activation causes dystrophinopathy in two Chinese families. European Journal of Human Genetics, 2020, 28, 947-955.	1.4	9
29	Base editing-mediated splicing correction therapy for spinal muscular atrophy. Cell Research, 2020, 30, 548-550.	5.7	33
30	Ataxic Severity Is Positively Correlated With Fatigue in Spinocerebellar Ataxia Type 3 Patients. Frontiers in Neurology, 2020, 11, 266.	1.1	8
31	Six Visual Rating Scales as A Biomarker for Monitoring Atrophied Brain Volume in Parkinson's Disease. , 2020, 11, 1082.		7
32	ATP1A1mutations cause intermediate Charcotâ€Marieâ€Tooth disease. Human Mutation, 2019, 40, 2334-2343.	1.1	11
33	Analysis of Exon Dosage Using Multiplex Ligation-Dependent Probe Amplification in Chinese Patients with Early-Onset Parkinson's Disease. European Neurology, 2019, 81, 246-253.	0.6	7
34	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.	0.9	14
35	Chinese patients with adrenoleukodystrophy and Zellweger spectrum disorder presenting with hereditary spastic paraplegia. Parkinsonism and Related Disorders, 2019, 65, 256-260.	1.1	9
36	The influence of initial symptoms on phenotypes in spinocerebellar ataxia type 3. Molecular Genetics & Lamp; Genomic Medicine, 2019, 7, e00719.	0.6	6

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37	Prognostic analysis of amyotrophic lateral sclerosis based on clinical features and plasma surfaceâ€enhanced Raman spectroscopy. Journal of Biophotonics, 2019, 12, e201900012.	1.1	13
38	Identification of <i>SLC20A2</i> deletions in patients with primary familial brain calcification. Clinical Genetics, 2019, 96, 53-60.	1.0	12
39	Spectrum of <i>SLC20A2</i> , <i>PDGFRB</i> , <i>PDGFB</i> , and <i>XPR1</i> mutations in a large cohort of patients with primary familial brain calcification. Human Mutation, 2019, 40, 392-403.	1.1	26
40	Generation of an integration-free induced pluripotent stem cell line, FJMUi001-A, from a hereditary spastic paraplegia patient carrying compound heterozygous p.P498L and p.R618W mutations in CAPN1 (SPG76). Stem Cell Research, 2019, 34, 101354.	0.3	7
41	Associations between neuroanatomical abnormality and motor symptoms in paroxysmal kinesigenic dyskinesia. Parkinsonism and Related Disorders, 2019, 62, 134-140.	1.1	17
42	Holmes tremor with impairment of the Guillain-Mollaret triangle following medullar hemorrhage. Neurological Sciences, 2018, 39, 1305-1306.	0.9	3
43	High frequency of the TARDBP p.M337ÂV mutation among south-eastern Chinese patients with familial amyotrophic lateral sclerosis. BMC Neurology, 2018, 18, 35.	0.8	6
44	Identification of a Novel Homozygous Splice-Site Mutation in SCARB2 that Causes Progressive Myoclonus Epilepsy with or without Renal Failure. Chinese Medical Journal, 2018, 131, 1575-1583.	0.9	3
45	A "Triple Trouble―Case of Facioscapulohumeral Muscular Dystrophy Accompanied by Peripheral Neuropathy and Myoclonic Epilepsy. Chinese Medical Journal, 2018, 131, 2164-2171.	0.9	2
46	Clinical and genetic investigation in Chinese patients with demyelinating Charcotâ€Marieâ€Tooth disease. Journal of the Peripheral Nervous System, 2018, 23, 216-226.	1.4	12
47	Biallelic Mutations in MYORG Cause Autosomal Recessive Primary Familial Brain Calcification. Neuron, 2018, 98, 1116-1123.e5.	3.8	111
48	Clinical features of familial amyloid polyneuropathy carrying transthyretin mutations in four Chinese kindreds. Journal of the Peripheral Nervous System, 2017, 22, 19-26.	1.4	16
49	Application of urine cells in drug intervention for spinal muscular atrophy. Experimental and Therapeutic Medicine, 2017, 14, 1993-1998.	0.8	3
50	Clinical features and mutation spectrum in Chinese patients with <scp>CADASIL</scp> : A multicenter retrospective study. CNS Neuroscience and Therapeutics, 2017, 23, 707-716.	1.9	41
51	Analysis of gene expression and functional characterization of XPR1: a pathogenic gene for primary familial brain calcification. Cell and Tissue Research, 2017, 370, 267-273.	1.5	21
52	Mutation screening of PDGFB gene in Chinese population with primary familial brain calcification. Gene, 2017, 597, 17-22.	1.0	8
53	Modeling the differential phenotypes of spinal muscular atrophy with high-yield generation of motor neurons from human induced pluripotent stem cells. Oncotarget, 2017, 8, 42030-42042.	0.8	17
54	Modeling the phenotype of spinal muscular atrophy by the direct conversion of human fibroblasts to motor neurons. Oncotarget, 2017, 8, 10945-10953.	0.8	20

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55	A Historical Cohort Study on the Efficacy of Glucocorticoids and Riboflavin Among Patients with Late-onset Multiple Acyl-CoA Dehydrogenase Deficiency. Chinese Medical Journal, 2016, 129, 142-146.	0.9	17
56	Skeletal Muscle Magnetic Resonance Imaging of the Lower Limbs in Late-onset Lipid Storage Myopathy with Electron Transfer Flavoprotein Dehydrogenase Gene Mutations. Chinese Medical Journal, 2016, 129, 1425-1431.	0.9	12
57	Serum Cytokines Th1, Th2, and Th17 Expression Profiling in Active Lupus Nephritis-IV: From a Southern Chinese Han Population. Mediators of Inflammation, 2016, 2016, 1-10.	1.4	31
58	Spectrum and Classification of <i>ATP7B</i> Variants in a Large Cohort of Chinese Patients with Wilson's Disease Guides Genetic Diagnosis. Theranostics, 2016, 6, 638-649.	4.6	71
59	Rivastigmine Patch in Chinese Patients with Probable Alzheimer's disease: A 24â€week, Randomized, Doubleâ€Blind Parallelâ€Group Study Comparing Rivastigmine Patch (9.5 mg/24 h) with Capsule (6 mg Twice) T	j ETiQq1 1	0. <b>78</b> 4314 rg
60	Mitochondrial <scp>NADH</scp> 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.	1.9	15
61	Neuroglobin Overexpression Inhibits AMPK Signaling and Promotes Cell Anabolism. Molecular Neurobiology, 2016, 53, 1254-1265.	1.9	18
62	Risk factors associated with acute/subacute cerebral infarction in HIV-negative patients with cryptococcal meningitis. Journal of the Neurological Sciences, 2016, 364, 19-23.	0.3	7
63	Significant clinical heterogeneity with similar ETFDH genotype in three Chinese patients with late-onset multiple acyl-CoA dehydrogenase deficiency. Neurological Sciences, 2016, 37, 1099-1105.	0.9	27
64	Large Animal Stroke Models vs. Rodent Stroke Models, Pros and Cons, and Combination?. Acta Neurochirurgica Supplementum, 2016, 121, 77-81.	0.5	15
65	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.	1.1	27
66	Mitochondrial DNA Haplogroups and the Risk of Sporadic Parkinson's Disease in Han Chinese. Chinese Medical Journal, 2015, 128, 1748-1754.	0.9	16
67	New Insights into Genotype-phenotype Correlations in Chinese Facioscapulohumeral Muscular Dystrophy. Chinese Medical Journal, 2015, 128, 1707-1713.	0.9	26
68	Intermediate-length polyglutamine in ATXN2 is a possible risk factor among Eastern Chinese patients with amyotrophic lateral sclerosis. Neurobiology of Aging, 2015, 36, 1603.e11-1603.e14.	1.5	13
69	Growth Hormone Deficiency in a Dopa-Responsive Dystonia Patient With a Novel Mutation of Guanosine Triphosphate Cyclohydrolase 1 Gene. Journal of Child Neurology, 2015, 30, 796-799.	0.7	4
70	Quantity of Cerebral Microbleeds, Antiplatelet Therapy, and Intracerebral Hemorrhage Outcomes: A Systematic Review and Meta-analysis. Journal of Stroke and Cerebrovascular Diseases, 2015, 24, 2728-2737.	0.7	39
71	Genotype-Phenotype Correlation in Chinese Patients with Spinal and Bulbar Muscular Atrophy. PLoS ONE, 2015, 10, e0122279.	1.1	17
72	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.	1.1	11

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73	Variations of <i>IGHMBP2 </i> Variations of <i>IGHMBP2  Iournal of Child Neurology, 2014, 29, NP35-NP39.</i>	0.7	4
74	Novel SLC20A2 mutations identified in southern Chinese patients with idiopathic basal ganglia calcification. Gene, 2013, 529, 159-162.	1.0	42
75	Expression and activity of the TLR4/NF-ΰB signaling pathway in mouse intestine following administration of a short-term high-fat diet. Experimental and Therapeutic Medicine, 2013, 6, 635-640.	0.8	37
76	The R219K polymorphism in the ATP-binding cassette transporter 1 gene has a protective effect on atherothrombotic cerebral infarction in Chinese Han ethnic population. Neurobiology of Aging, 2010, 31, 647-653.	1.5	12