

# Fang Fang

## List of Publications by Year in descending order

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110  
papers

1,598  
citations

430874

18  
h-index

395702

33  
g-index

117  
all docs

117  
docs citations

117  
times ranked

2503  
citing authors

#	ARTICLE	IF	CITATIONS
1	Whole genome and exome sequencing identify <i>NDUFV2</i> mutations as a new cause of progressive cavitating leukoencephalopathy. <i>Journal of Medical Genetics</i> , 2022, 59, 351-357.	3.2	5
2	Phenotypes and genotypes of mitochondrial diseases with mtDNA variations in Chinese children: A multi-center study. <i>Mitochondrion</i> , 2022, 62, 139-150.	3.4	6
3	Generation of an iPSC line from a patient with early-onset epileptic encephalopathy carrying <i>CARS2</i> (p.G476R) mutation. <i>Stem Cell Research</i> , 2022, 59, 102633.	0.7	0
4	Olfactory dysfunction is associated with cognitive impairment in patients with obstructive sleep apnea: a cross-sectional study. <i>European Archives of Oto-Rhino-Laryngology</i> , 2022, 279, 1979-1987.	1.6	3
5	Leigh Syndrome: A Study of 209 Patients at the Beijing Children's Hospital. <i>Annals of Neurology</i> , 2022, 91, 466-482.	5.3	10
6	Identification and characterization of novel <i>MPC1</i> gene variants causing mitochondrial pyruvate carrier deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2022, 45, 264-277.	3.6	7
7	The association between glucocorticoid receptor ( <i>NR3C1</i> ) gene polymorphism and difficult-to-treat rhinosinusitis. <i>European Archives of Oto-Rhino-Laryngology</i> , 2022, 279, 3981-3987.	1.6	3
8	Genetic analysis and clinical significance of a rare t(1;12)(q21;p13) in a patient with high-risk myelodysplastic syndrome. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2022, , e1893.	1.2	1
9	Chinese patients with p.Arg756 mutations of <i>ATP1A3</i> : Clinical manifestations, treatment, and follow-up. <i>Pediatric Investigation</i> , 2022, 6, 5-10.	1.4	2
10	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. <i>Genome Medicine</i> , 2022, 14, 38.	8.2	85
11	Phenotypic and Genotypic Characteristics of <i>SCN1A</i> Associated Seizure Diseases. <i>Frontiers in Molecular Neuroscience</i> , 2022, 15, 821012.	2.9	5
12	Clinical analysis of <i>CHD2</i> gene mutations in pediatric patients with epilepsy. <i>Pediatric Investigation</i> , 2022, 6, 93-99.	1.4	3
13	Novel Loss-of-Function Variants in <i>CHD2</i> Cause Childhood-Onset Epileptic Encephalopathy in Chinese Patients. <i>Genes</i> , 2022, 13, 908.	2.4	1
14	Cerebral small vessel disease caused by <i>PLOD3</i> mutation: Expanding the phenotypic spectrum of lysyl hydroxylase deficiency. <i>Pediatric Investigation</i> , 2022, 6, 219-223.	1.4	3
15	Cost-Effectiveness of Evolocumab Therapy for Myocardial Infarction: The Chinese Healthcare Perspective. <i>Cardiovascular Drugs and Therapy</i> , 2021, 35, 775-785.	2.6	7
16	Low arousal threshold is associated with unfavorable shift of PAP compliance over time in patients with OSA. <i>Sleep and Breathing</i> , 2021, 25, 887-895.	1.7	5
17	Pediatric Leigh Syndrome: Neuroimaging Features and Genetic Correlations. <i>Annals of Neurology</i> , 2021, 89, 629-631.	5.3	4
18	Quantification of myocardial deformation in patients with Fabry disease by cardiovascular magnetic resonance feature tracking imaging. <i>Cardiovascular Diagnosis and Therapy</i> , 2021, 11, 91-101.	1.7	8

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19	Novel truncating mutations in ASXL1 identified in two boys with Bohring-Opitz syndrome. <i>European Journal of Medical Genetics</i> , 2021, 64, 104155.	1.3	2
20	Clinical, Metabolic, and Genetic Analysis and Follow-Up of Eight Patients With HIBCH Mutations Presenting With Leigh/Leigh-Like Syndrome. <i>Frontiers in Pharmacology</i> , 2021, 12, 605803.	3.5	4
21	Whole exome sequencing identifies a novel homozygous MECP2 mutation in a Chinese patient with childhood-onset dystonia and basal ganglia abnormalities, without optic atrophy. <i>Mitochondrion</i> , 2021, 57, 222-229.	3.4	13
22	Efficacy of the ketogenic diet on ACTH-resistant infantile spasm: a multicentre prospective control study. <i>Epileptic Disorders</i> , 2021, 23, 337-345.	1.3	5
23	Case Report: Autoimmune Encephalitis Associated With Anti-glutamic Acid Decarboxylase Antibodies: A Pediatric Case Series. <i>Frontiers in Neurology</i> , 2021, 12, 641024.	2.4	9
24	Cost-Effectiveness of Alirocumab for the Secondary Prevention of Cardiovascular Events after Myocardial Infarction in the Chinese Setting. <i>Frontiers in Pharmacology</i> , 2021, 12, 648244.	3.5	5
25	Analytical validation of GMEX rapid point-of-care CYP2C19 genotyping system for the CHANCE-2 trial. <i>Stroke and Vascular Neurology</i> , 2021, 6, 274-279.	3.3	6
26	A novel homozygous mutation in ATP13A2 gene causing pure hereditary spastic paraplegia. <i>Parkinsonism and Related Disorders</i> , 2021, 86, 58-60.	2.2	2
27	Identification of a Novel Variant in MT-CO3 Causing MELAS. <i>Frontiers in Genetics</i> , 2021, 12, 638749.	2.3	2
28	Case Report: Clinical Features of Childhood Leukoencephalopathy With Cerebral Calcifications and Cysts Due to SNORD118 Variants. <i>Frontiers in Neurology</i> , 2021, 12, 585606.	2.4	2
29	Clinical Features and Outcomes of Anti-N-Methyl-d-Aspartate Receptor Encephalitis in Infants and Toddlers. <i>Pediatric Neurology</i> , 2021, 119, 27-33.	2.1	5
30	Report of the Largest Chinese Cohort With SLC19A3 Gene Defect and Literature Review. <i>Frontiers in Genetics</i> , 2021, 12, 683255.	2.3	10
31	Novel Mutations in the GTPBP3 Gene for Mitochondrial Disease and Characteristics of Related Phenotypic Spectrum: The First Three Cases From China. <i>Frontiers in Genetics</i> , 2021, 12, 611226.	2.3	6
32	Association Between OSA and Quantitative Atherosclerotic Plaque Burden. <i>Chest</i> , 2021, 160, 1864-1874.	0.8	15
33	Biallelic COA7-Variants Leading to Developmental Regression With Progressive Spasticity and Brain Atrophy in a Chinese Patient. <i>Frontiers in Genetics</i> , 2021, 12, 685035.	2.3	7
34	Automatic detection of interictal ripples on scalp EEG to evaluate the effect and prognosis of ACTH therapy in patients with infantile spasms. <i>Epilepsia</i> , 2021, 62, 2240-2251.	5.1	14
35	HPDL deficiency causes a neuromuscular disease by impairing the mitochondrial respiration. <i>Journal of Genetics and Genomics</i> , 2021, 48, 727-736.	3.9	5
36	Age-dependent characteristics and prognostic factors of pediatric anti-N-methyl-d-aspartate receptor encephalitis in a Chinese single-center study. <i>European Journal of Paediatric Neurology</i> , 2021, 34, 67-73.	1.6	5

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37	Dominant <i>KPNA3</i> Mutations Cause Infantile-Onset Hereditary Spastic Paraplegia. <i>Annals of Neurology</i> , 2021, 90, 738-750.	5.3	5
38	Identification of a Novel m.3955G>A Variant in MT-ND1 Associated with Leigh Syndrome. <i>Mitochondrion</i> , 2021, 62, 13-23.	3.4	3
39	Decreased nasal nitric oxide levels: A potential marker of decreased olfactory discrimination in chronic rhinosinusitis. <i>Journal of Laryngology and Otology</i> , 2021, , 1-28.	0.8	0
40	Clinical Attributes and Electroencephalogram Analysis of Patients With Varying Alpers Syndrome Genotypes. <i>Frontiers in Pharmacology</i> , 2021, 12, 669516.	3.5	1
41	Recovery of Gonadal Hormone Level Is a Potential Marker for the Response and Prognosis in POEMS Syndrome Patients Treated with Bortezomib Based Combined Chemotherapy. <i>Blood</i> , 2021, 138, 4741-4741.	1.4	0
42	Independent Role of Nasal Congestion in Positive Airway Pressure Compliance for OSA Treatment. <i>Otolaryngology - Head and Neck Surgery</i> , 2021, , 019459982110645.	1.9	2
43	Transcranial direct current stimulation reduces seizure frequency in patients with refractory focal epilepsy: A randomized, double-blind, sham-controlled, and three-arm parallel multicenter study. <i>Brain Stimulation</i> , 2020, 13, 109-116.	1.6	70
44	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. <i>Brain</i> , 2020, 143, e8-e8.	7.6	18
45	Usefulness of Cathepsin S to Predict Risk for Obstructive Sleep Apnea among Patients with Type 2 Diabetes. <i>Disease Markers</i> , 2020, 2020, 1-8.	1.3	3
46	EMMPRIN: A potential biomarker for predicting the presence of obstructive sleep apnea. <i>Clinica Chimica Acta</i> , 2020, 510, 317-322.	1.1	4
47	Novel ECHS1 mutations in Leigh syndrome identified by whole-exome sequencing in five Chinese families: case report. <i>BMC Medical Genetics</i> , 2020, 21, 149.	2.1	10
48	Case Report: Rapid Treatment of Uridine-Responsive Epileptic Encephalopathy Caused by CAD Deficiency. <i>Frontiers in Pharmacology</i> , 2020, 11, 608737.	3.5	8
49	Current Status, Diagnosis, and Treatment Recommendation for Tic Disorders in China. <i>Frontiers in Psychiatry</i> , 2020, 11, 774.	2.6	27
50	Obstructive sleep apnoea and inflammation in age-dependent cardiovascular disease. <i>European Heart Journal</i> , 2020, 41, 2503-2503.	2.2	6
51	Efficacy and safety of corticosteroids in COVID-19 based on evidence for COVID-19, other coronavirus infections, influenza, community-acquired pneumonia and acute respiratory distress syndrome: a systematic review and meta-analysis. <i>Cmaj</i> , 2020, 192, E756-E767.	2.0	166
52	CPAP is associated with decreased risk of AF recurrence in patients with OSA, especially those younger and slimmer: a meta-analysis. <i>Journal of Interventional Cardiac Electrophysiology</i> , 2020, 58, 369-379.	1.3	13
53	The association between circulating APRIL levels and severity of obstructive sleep apnea in Chinese adults. <i>Clinica Chimica Acta</i> , 2020, 508, 161-169.	1.1	4
54	Gene panel for Mendelian strokes. <i>Stroke and Vascular Neurology</i> , 2020, 5, 416-421.	3.3	12

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55	Clinical phenotypes, genotypes and treatment in Chinese dystonia patients with KMT2B variants. <i>Parkinsonism and Related Disorders</i> , 2020, 77, 76-82.	2.2	13
56	The impact of obstructive apnea sleep syndrome on chemical function. <i>Sleep and Breathing</i> , 2020, 24, 1549-1555.	1.7	6
57	Resective epilepsy surgery in tuberous sclerosis complex: a nationwide multicentre retrospective study from China. <i>Brain</i> , 2020, 143, 570-581.	7.6	55
58	Cardiac cycle time intervals are back again. <i>International Journal of Cardiology</i> , 2020, 312, 87-88.	1.7	0
59	Long-term efficacy of mycophenolate mofetil in myelin oligodendrocyte glycoprotein antibody-associated disorders. <i>Neurology: Neuroimmunology and NeuroInflammation</i> , 2020, 7, .	6.0	46
60	KLF10 Deficiency in CD4+ T Cells Triggers Obesity, Insulin Resistance, and Fatty Liver. <i>Cell Reports</i> , 2020, 33, 108550.	6.4	30
61	A Symptomatic Female Patient with Duchenne Muscular Dystrophy Gene Mutation Showing Rimmed Vacuoles in Muscle Biopsy. <i>Neurology India</i> , 2020, 68, 518.	0.4	0
62	Vaccine-Associated Paralytic Poliomyelitis “ 8 PLADs, China, October 2012”–“March 2014. <i>China CDC Weekly</i> , 2020, 2, 955-961.	2.3	1
63	ESM promotes adhesion between monocytes and endothelial cells under intermittent hypoxia. <i>Journal of Cellular Physiology</i> , 2019, 234, 1512-1521.	4.1	43
64	A novel DDC gene deletion mutation in two Chinese mainland siblings with aromatic l-amino acid decarboxylase deficiency. <i>Brain and Development</i> , 2019, 41, 205-209.	1.1	10
65	Effect of uvulopalatopharyngoplasty (UPPP) on atherosclerosis and cardiac functioning in obstructive sleep apnea patients. <i>Acta Oto-Laryngologica</i> , 2019, 139, 793-797.	0.9	11
66	Circulating ESM-1 levels are correlated with the presence of coronary artery disease in patients with obstructive sleep apnea. <i>Respiratory Research</i> , 2019, 20, 188.	3.6	11
67	The Efficacy of Ketogenic Diet in 60 Chinese Patients With Dravet Syndrome. <i>Frontiers in Neurology</i> , 2019, 10, 625.	2.4	24
68	Two Chinese siblings with two novel KCTD7 mutations have dystonia or seizures and epileptic discharge on electroencephalograms. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2019, 70, 27-29.	2.0	4
69	Next-Generation Sequencing Analysis Reveals Novel Pathogenic Variants in Four Chinese Siblings With Late-Infantile Neuronal Ceroid Lipofuscinosis. <i>Frontiers in Genetics</i> , 2019, 10, 370.	2.3	10
70	Transcatheter versus surgical aortic valve replacement in low and intermediate risk patients with severe aortic stenosis: systematic review and meta-analysis of randomized controlled trials and propensity score matching observational studies. <i>Journal of Thoracic Disease</i> , 2019, 11, 1945-1962.	1.4	20
71	Neurochondrin Antibody Serum Positivity in Three Cases of Autoimmune Cerebellar Ataxia. <i>Cerebellum</i> , 2019, 18, 1137-1142.	2.5	22
72	O259 Reduction of the Apnea-Hypopnea Duration Ameliorates Endothelial Dysfunction, Vascular Inflammation, and Systemic Hypertension in a Rat Model of Obstructive Sleep Apnea. <i>Sleep</i> , 2019, 42, A106-A106.	1.1	0

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73	Compound Heterozygous CHAT Gene Mutations of a Large Deletion and a Missense Variant in a Chinese Patient With Severe Congenital Myasthenic Syndrome With Episodic Apnea. <i>Frontiers in Pharmacology</i> , 2019, 10, 259.	3.5	9
74	The reduction of apnea-hypopnea duration ameliorates endothelial dysfunction, vascular inflammation, and systemic hypertension in a rat model of obstructive sleep apnea. <i>Sleep and Breathing</i> , 2019, 23, 1187-1196.	1.7	11
75	Clinical Assessments and EEG Analyses of Encephalopathies Associated With Dynamin-1 Mutation. <i>Frontiers in Pharmacology</i> , 2019, 10, 1454.	3.5	11
76	0575 Validation of a Portable Monitoring for the Diagnosis of Obstructive Sleep Apnea: Electrocardiogram-based Cardiopulmonary Coupling. <i>Sleep</i> , 2019, 42, A229-A229.	1.1	0
77	An inherited KMT2B duplication variant in a Chinese family with dystonia and/or development delay. <i>Parkinsonism and Related Disorders</i> , 2019, 63, 227-228.	2.2	15
78	Lack of association between valproic acid response and polymorphisms of its metabolism, transport, and receptor genes in children with focal seizures. <i>Neurological Sciences</i> , 2019, 40, 523-528.	1.9	3
79	TNFRSF11B: A potential plasma biomarker for diagnosis of obstructive sleep apnea. <i>Clinica Chimica Acta</i> , 2019, 490, 39-45.	1.1	13
80	CYP2C19 genotype and adverse cardiovascular outcomes after stent implantation in clopidogrel-treated Asian populations: A systematic review and meta-analysis. <i>Platelets</i> , 2019, 30, 229-240.	2.3	42
81	The contribution of chronic intermittent hypoxia to OSAHS: From the perspective of serum extracellular microvesicle proteins. <i>Metabolism: Clinical and Experimental</i> , 2018, 85, 97-108.	3.4	23
82	Effect of CYP2C19, UGT1A8, and UGT2B7 on valproic acid clearance in children with epilepsy: a population pharmacokinetic model. <i>European Journal of Clinical Pharmacology</i> , 2018, 74, 1029-1036.	1.9	19
83	Effects of UGT2B7, SCN1A and CYP3A4 on the therapeutic response of sodium valproate treatment in children with generalized seizures. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2018, 58, 96-100.	2.0	18
84	Incremental prognostic value of multichamber deformation imaging and renal function status to predict adverse outcome in heart failure with reduced ejection fraction. <i>Echocardiography</i> , 2018, 35, 450-458.	0.9	10
85	Comparison of myelin oligodendrocyte glycoprotein (MOG)-antibody disease and AQP4-IgG-positive neuromyelitis optica spectrum disorder (NMOSD) when they co-exist with anti-NMDA (N-methyl-D-aspartate) receptor encephalitis. <i>Multiple Sclerosis and Related Disorders</i> , 2018, 20, 144-152.	2.0	89
86	The fallacy of resting echocardiographic parameters of cardiac function in heart failure with preserved ejection fraction. <i>European Journal of Heart Failure</i> , 2018, 20, 619-619.	7.1	2
87	A Retrospective Study to Compare the Use of the Mean Apnea-Hypopnea Duration and the Apnea-Hypopnea Index with Blood Oxygenation and Sleep Patterns in Patients with Obstructive Sleep Apnea Diagnosed by Polysomnography. <i>Medical Science Monitor</i> , 2018, 24, 1887-1893.	1.1	19
88	Bi-allelic ADPRHL2 Mutations Cause Neurodegeneration with Developmental Delay, Ataxia, and Axonal Neuropathy. <i>American Journal of Human Genetics</i> , 2018, 103, 817-825.	6.2	40
89	Exogenous hydrogen sulfide ameliorates high glucose-induced myocardial injury & inflammation via the C1RP-MAPK signaling pathway in H9c2 cardiac cells. <i>Life Sciences</i> , 2018, 208, 315-324.	4.3	22
90	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 120.	2.7	61

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91	Does Masked Hypertension Cause Early Left Ventricular Impairment in Youth?. <i>Frontiers in Pediatrics</i> , 2018, 6, 167.	1.9	8
92	Establishing age and sex dependent upper reference limits for the plasma lipoprotein (a) in a Chinese health check-up population and according to its relative risk of primary myocardial infarction. <i>Clinica Chimica Acta</i> , 2018, 484, 232-236.	1.1	15
93	Phenotype-Driven Virtual Panel Is an Effective Method to Analyze WES Data of Neurological Disease. <i>Frontiers in Pharmacology</i> , 2018, 9, 1529.	3.5	15
94	DHX32 expression is an indicator of poor breast cancer prognosis. <i>Oncology Letters</i> , 2017, 13, 942-948.	1.8	5
95	Molecular spectrum of excision repair cross-complementation group 8 gene defects in Chinese patients with Cockayne syndrome type A. <i>Scientific Reports</i> , 2017, 7, 13686.	3.3	7
96	Genetic polymorphisms and valproic acid plasma concentration in children with epilepsy on valproic acid monotherapy. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2017, 51, 22-26.	2.0	17
97	The clinical and genetic characteristics in children with mitochondrial disease in China. <i>Science China Life Sciences</i> , 2017, 60, 746-757.	4.9	32
98	Automated left heart chamber volumetric assessment using three-dimensional echocardiography in Chinese adolescents. <i>Journal of Animal Science and Technology</i> , 2017, 4, 53-61.	2.5	10
99	High expression of UBE2C is associated with the aggressive progression and poor outcome of malignant glioma. <i>Oncology Letters</i> , 2016, 11, 2300-2304.	1.8	42
100	In-stent restenosis in a polytetrafluoroethylene covered stent combined with drug eluting stents: potential pathogenesis revealed by optical coherence tomography. <i>International Journal of Cardiology</i> , 2015, 198, 42-44.	1.7	2
101	The patient's selection of PARACHUTE® endoventricular partitioning device: The important role of detailed echocardiography. <i>International Journal of Cardiology</i> , 2015, 195, 176-179.	1.7	2
102	Predictors of mid-term functional tricuspid regurgitation after device closure of atrial septal defect in adults: Impact of pre-operative tricuspid valve remodeling. <i>International Journal of Cardiology</i> , 2015, 187, 447-452.	1.7	13
103	The diagnostic value of cerebrospinal fluids procalcitonin and lactate for the differential diagnosis of post-neurosurgical bacterial meningitis and aseptic meningitis. <i>Clinical Biochemistry</i> , 2015, 48, 50-54.	1.9	57
104	Left anterior descending coronary artery flow impaired by right ventricular apical pacing: The role of systolic dyssynchrony. <i>International Journal of Cardiology</i> , 2014, 176, 80-85.	1.7	6
105	Seizure Control of Current Shunt on Rats with Temporal Lobe Epilepsy and Neocortical Epilepsy. <i>PLoS ONE</i> , 2014, 9, e86477.	2.5	2
106	Variation in right ventricular volumes assessment by real-time three-dimensional echocardiography between dilated and normal right ventricle: Comparison with cardiac magnetic resonance imaging. <i>International Journal of Cardiology</i> , 2013, 168, 4391-4393.	1.7	8
107	Deciphering the Mysteries of Crisscross Heart by Transthoracic Echocardiography. <i>Echocardiography</i> , 2011, 28, 104-108.	0.9	8
108	An Unusual Cause of Right Heart Failure in a Patient With Previous Hysterectomy. <i>Journal of Ultrasound in Medicine</i> , 2010, 29, 1647-1650.	1.7	2

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109	Identification of Unusual Conditions after Atrial Septal Defect Repair by Systematic Transthoracic Echocardiographic Assessment. Echocardiography, 2008, 25, 1094-1100.	0.9	3
110	A 5-year-old child presenting with tumor-like primary angiitis of the central nervous system. Pediatric Investigation, 0, , .	1.4	0