

Jie-Yuan Jin

List of Publications by Year in descending order

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papers

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#	ARTICLE	IF	CITATIONS
1	Increased Reticulon 3 (RTN3) Leads to Obesity and Hypertriglyceridemia by Interacting With Heat Shock Protein Family A (Hsp70) Member 5 (HSPA5). <i>Circulation</i> , 2018, 138, 1828-1838.	1.6	26
2	Whole exome sequencing identifies a novel mutation (c.333A>T) of TNNI3K in a Chinese family with dilated cardiomyopathy and cardiac conduction disease. <i>Gene</i> , 2018, 648, 63-67.	2.2	25
3	The genetic spectrum of familial hypercholesterolemia in the central south region of China. <i>Atherosclerosis</i> , 2017, 258, 84-88.	0.8	22
4	A <i>de novo</i> mutation of <i>SMYD1</i> (p.F272L) is responsible for hypertrophic cardiomyopathy in a Chinese patient. <i>Clinical Chemistry and Laboratory Medicine</i> , 2019, 57, 532-539.	2.3	19
5	Whole-exome sequencing identifies a novel mutation of <i>GPD1L</i> (R189X) associated with familial conduction disease and sudden death. <i>Journal of Cellular and Molecular Medicine</i> , 2018, 22, 1350-1354.	3.6	12
6	Novel Compound Heterozygous DST Variants Causing Hereditary Sensory and Autonomic Neuropathies VI in Twins of a Chinese Family. <i>Frontiers in Genetics</i> , 2020, 11, 492.	2.3	10
7	A Novel Nonsense Mutation of ABCA8 in a Han-Chinese Family With ASCVD Leads to the Reduction of HDL-c Levels. <i>Frontiers in Genetics</i> , 2020, 11, 755.	2.3	9
8	Exome sequencing identifies a novel nonsense mutation of Ring Finger Protein 207 in a Chinese family with Long QT syndrome and syncope. <i>Journal of Human Genetics</i> , 2019, 64, 233-238.	2.3	8
9	A novel heterozygous variant p.(Trp538Arg) of SYNM is identified by whole-exome sequencing in a Chinese family with dilated cardiomyopathy. <i>Annals of Human Genetics</i> , 2019, 83, 95-99.	0.8	7
10	The Novel Compound Heterozygous Mutations of <i>ECEL1</i> Identified in a Family with Distal Arthrogryposis Type 5D. <i>BioMed Research International</i> , 2020, 2020, 1-6.	1.9	7
11	Whole-Exome Sequencing Identifies a Novel TRPM4 Mutation in a Chinese Family with Atrioventricular Block. <i>BioMed Research International</i> , 2021, 2021, 1-6.	1.9	7
12	A novel mutation (c.1010G>T; p.R337L) in <i>TP63</i> as a cause of split-hand/foot malformation with hypodontia. <i>Journal of Gene Medicine</i> , 2019, 21, e3122.	2.8	6
13	Loss of RTN3 phenocopies chronic kidney disease and results in activation of the IGF2-JAK2 pathway in proximal tubular epithelial cells. <i>Experimental and Molecular Medicine</i> , 2022, 54, 653-661.	7.7	5
14	Whole-exome sequencing identifies a Novel <i>SCN5A</i> mutation (C335R) in a Chinese family with arrhythmia. <i>Cardiology in the Young</i> , 2018, 28, 688-691.	0.8	4
15	Whole-exome sequencing reveals doubly novel heterozygous <i>Myosin Binding Protein C</i> and <i>Titin</i> mutations in a Chinese patient with severe dilated cardiomyopathy. <i>Cardiology in the Young</i> , 2018, 28, 1410-1414.	0.8	4
16	GLIS Family Zinc Finger 1 was First Linked With Preaxial Polydactyly I in Humans by Stepwise Genetic Analysis. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 781388.	3.7	4
17	Whole Exome Sequencing Identified a 13 Base Pair Deletion-mutation in a Patient with Restrictive Cardiomyopathy and Left Ventricle Hypertrophy. <i>Annals of Clinical and Laboratory Science</i> , 2019, 49, 838-840.	0.2	4
18	A Novel COMP Mutated Allele Identified in a Chinese Family with Pseudoachondroplasia. <i>BioMed Research International</i> , 2021, 2021, 1-8.	1.9	3

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19	A mutation of beta-tropomyosin gene in a Chinese family with distal arthrogyriposis type I. <i>International Journal of Clinical and Experimental Pathology</i> , 2017, 10, 11137-11142.	0.5	3
20	A novel splice-site mutation of WRN (c.IVS28+2T>C) identified in a consanguineous family with Werner Syndrome. <i>Molecular Medicine Reports</i> , 2017, 15, 3735-3738.	2.4	2
21	Microduplication of 10q26.3 in a Chinese hypertriglyceridemia patient. <i>Molecular and Cellular Probes</i> , 2018, 37, 28-31.	2.1	2
22	A novel proximal 3q29 chromosome microdeletion in a Chinese patient with Chiari malformation type II and Sprengel's deformity. <i>Molecular Cytogenetics</i> , 2018, 11, 8.	0.9	2
23	Identification of a Novel Variant of ARHGAP29 in a Chinese Family with Nonsyndromic Cleft Lip and Palate. <i>BioMed Research International</i> , 2020, 2020, 1-6.	1.9	2
24	Case Report: A Novel Gross Deletion in PAX3 (10.26 kb) Identified in a Chinese Family With Waardenburg Syndrome by Third-Generation Sequencing. <i>Frontiers in Genetics</i> , 2021, 12, 705973.	2.3	2
25	Identification of an unknown frameshift variant of <i>NOG</i> in a Han Chinese family with proximal symphalangism. <i>Bioscience Reports</i> , 2020, 40, .	2.4	2
26	Compound heterozygous mutations cause mucopolidosis II or III alpha/beta in two Chinese families. <i>International Journal of Clinical and Experimental Pathology</i> , 2019, 12, 2981-2988.	0.5	2
27	Microduplications of 10q24 Detected in Two Chinese Patients with Split-hand/foot Malformation Type 3. <i>Annals of Clinical and Laboratory Science</i> , 2017, 47, 754-757.	0.2	2
28	Identification of a novel GATA binding protein 5 variant (c.830C>T/p.P277L) damaging the nuclear translocation and causing tetralogy of Fallot. <i>QJM - Monthly Journal of the Association of Physicians</i> , 2022, 115, 256-258.	0.5	2
29	Identification of a novel mutation in the C6 gene of a Han Chinese C6SD child with meningococcal disease. <i>Experimental and Therapeutic Medicine</i> , 2021, 21, 510.	1.8	1
30	Case Report: A Homozygous Mutation (p.Y62X) of Phospholipase D3 May Lead to a New Leukoencephalopathy Syndrome. <i>Frontiers in Aging Neuroscience</i> , 2021, 13, 671296.	3.4	1
31	A novel <i>POF1B</i> variant in a Chinese patient is associated with premature ovarian failure. <i>Clinical Genetics</i> , 2021, 100, 773-774.	2.0	1
32	Genetic analysis combined with 3D-printing assistant surgery in diagnosis and treatment for an X-linked hypophosphatemia patient. <i>Journal of Clinical Laboratory Analysis</i> , 2022, 36, e24243.	2.1	1
33	Case Report: Identification of the First Synonymous Variant of Myosin Binding Protein C3 (c.24A>C, Tj ETQq1 1 0.784314 rgBT /Ove Cardiovascular Medicine, 2022, 9, 806977.	2.4	1
34	Identification of a Novel Arginine Vasopressin Receptor 2 Mutation (p.V183M) in a Chinese Family with Nephrogenic Diabetes Insipidus. <i>Molecular Syndromology</i> , 2020, 11, 130-134.	0.8	0
35	Identification of Two Novel Frameshift Mutations in Exostosin 1 in Two Families with Multiple Osteochondromas. <i>Molecular Syndromology</i> , 2021, 12, 96-100.	0.8	0
36	Case Report: A Novel CACNA1S Mutation Associated With Hypokalemic Periodic Paralysis in a Chinese Family. <i>Frontiers in Genetics</i> , 2021, 12, 743184.	2.3	0

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37	Whole-exome sequencing identified a novel mutation of BMP2 in a Chinese family with pulmonary arterial hypertension. <i>International Journal of Transgender Health</i> , 2021, 14, 874-880.	2.3	0