Jie-Yuan Jin

List of Publications by Year
in descending order

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37	208	1307594	1199594
papers	citations	7 h-index	12 g-index

37	37	37	375
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Genetic analysis combined with 3Dâ€printing assistant surgery in diagnosis and treatment for an Xâ€linked hypophosphatemia patient. Journal of Clinical Laboratory Analysis, 2022, 36, e24243.	2.1	1
2	Case Report: Identification of the First Synonymous Variant of Myosin Binding Protein C3 (c.24A>C,) Tj ETQq0 0 0 rgBT /Overlock 10 Cardiovascular Medicine, 2022, 9, 806977.	2.4	1
3	Identification of a novel GATA binding protein 5 variant (c.830C>T/p.P277L) damaging the nuclear translocation and causing tetralogy of Fallot. QJM - Monthly Journal of the Association of Physicians, 2022, 115, 256-258.	0.5	2
4	Loss of RTN3 phenocopies chronic kidney disease and results in activation of the IGF2-JAK2 pathway in proximal tubular epithelial cells. Experimental and Molecular Medicine, 2022, 54, 653-661.	7.7	5
5	Identification of Two Novel Frameshift Mutations in Exostosin 1 in Two Families with Multiple Osteochondromas. Molecular Syndromology, 2021, 12, 96-100.	0.8	0
6	A Novel COMP Mutated Allele Identified in a Chinese Family with Pseudoachondroplasia. BioMed Research International, 2021, 2021, 1-8.	1.9	3
7	Identification of a novel mutation in the C6 gene of a Han Chinese C6SD child with meningococcal disease. Experimental and Therapeutic Medicine, 2021, 21, 510.	1.8	1
8	Whole-Exome Sequencing Identifies a Novel TRPM4 Mutation in a Chinese Family with Atrioventricular Block. BioMed Research International, 2021, 2021, 1-6.	1.9	7
9	Case Report: A Homozygous Mutation (p.Y62X) of Phospholipase D3 May Lead to a New Leukoencephalopathy Syndrome. Frontiers in Aging Neuroscience, 2021, 13, 671296.	3.4	1
10	Case Report: A Novel Gross Deletion in PAX3 (10.26 kb) Identified in a Chinese Family With Waardenburg Syndrome by Third-Generation Sequencing. Frontiers in Genetics, 2021, 12, 705973.	2.3	2
11	A novel POF1B variant in a Chinese patient is associated with premature ovarian failure. Clinical Genetics, 2021, 100, 773-774.	2.0	1
12	Case Report: A Novel CACNA1S Mutation Associated With Hypokalemic Periodic Paralysis in a Chinese Family. Frontiers in Genetics, 2021, 12, 743184.	2.3	0
13	Whole-exome sequencing identified a novel mutation of BMPR2 in a Chinese family with pulmonary arterial hypertension. International Journal of Transgender Health, 2021, 14, 874-880.	2.3	0
14	GLIS Family Zinc Finger 1 was First Linked With Preaxial Polydactyly I in Humans by Stepwise Genetic Analysis. Frontiers in Cell and Developmental Biology, 2021, 9, 781388.	3.7	4
15	A Novel Nonsense Mutation of ABCA8 in a Han-Chinese Family With ASCVD Leads to the Reduction of HDL-c Levels. Frontiers in Genetics, 2020, 11, 755.	2.3	9
16	Identification of a Novel Variant of ARHGAP29 in a Chinese Family with Nonsyndromic Cleft Lip and Palate. BioMed Research International, 2020, 2020, 1-6.	1.9	2
17	Novel Compound Heterozygous DST Variants Causing Hereditary Sensory and Autonomic Neuropathies VI in Twins of a Chinese Family. Frontiers in Genetics, 2020, 11, 492.	2.3	10
18	Identification of a Novel Arginine Vasopressin Receptor 2 Mutation (p.V183M) in a Chinese Family with Nephrogenic Diabetes Insipidus. Molecular Syndromology, 2020, 11, 130-134.	0.8	0

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19	The Novel Compound Heterozygous Mutations of ECEL1 Identified in a Family with Distal Arthrogryposis Type 5D. BioMed Research International, 2020, 2020, 1-6.	1.9	7
20	Identification of an unknown frameshift variant of NOG in a Han Chinese family with proximal symphalangism. Bioscience Reports, 2020, 40, .	2.4	2
21	A novel mutation (c.1010G>T; p.R337L) in TP63 as a cause of splitâ€hand/foot malformation with hypodontia. Journal of Gene Medicine, 2019, 21, e3122.	2.8	6
22	A de novo mutation of SMYD1 (p.F272L) is responsible for hypertrophic cardiomyopathy in a Chinese patient. Clinical Chemistry and Laboratory Medicine, 2019, 57, 532-539.	2.3	19
23	Exome sequencing identifies a novel nonsense mutation of Ring Finger Protein 207 in a Chinese family with Long QT syndrome and syncope. Journal of Human Genetics, 2019, 64, 233-238.	2.3	8
24	A novel heterozygous variant p.(Trp538Arg) of SYNM is identified by wholeâ€exome sequencing in a Chinese family with dilated cardiomyopathy. Annals of Human Genetics, 2019, 83, 95-99.	0.8	7
25	Compound heterozygous mutations cause mucolipidosis II or III alpha/beta in two Chinese families. International Journal of Clinical and Experimental Pathology, 2019, 12, 2981-2988.	0.5	2
26	Whole Exome Sequencing Identified a 13 Base Pair Deletion-mutation in a Patient with Restrictive Cardiomyopathy and Left Ventricle Hypertrophy. Annals of Clinical and Laboratory Science, 2019, 49, 838-840.	0.2	4
27	Wholeâ€exome sequencing identifies a novel mutation of GPD1L (R189X) associated with familial conduction disease and sudden death. Journal of Cellular and Molecular Medicine, 2018, 22, 1350-1354.	3.6	12
28	Whole exome sequencing identifies a novel mutation (c.333â€¯+â€¯2Tâ€¯>â€¯C) of TNNI3K in a Chinese family with dilated cardiomyopathy and cardiac conduction disease. Gene, 2018, 648, 63-67.	2.2	25
29	Whole-exome sequencing identifies a Novel SCN5A mutation (C335R) in a Chinese family with arrhythmia. Cardiology in the Young, 2018, 28, 688-691.	0.8	4
30	Microduplication of 10q26.3 in a Chinese hypertriglyceridemia patient. Molecular and Cellular Probes, 2018, 37, 28-31.	2.1	2
31	Increased Reticulon 3 (RTN3) Leads to Obesity and Hypertriglyceridemia by Interacting With Heat Shock Protein Family A (Hsp70) Member 5 (HSPA5). Circulation, 2018, 138, 1828-1838.	1.6	26
32	A novel proximal 3q29 chromosome microdeletion in a Chinese patient with Chiari malformation type II and Sprengelâ€™s deformity. Molecular Cytogenetics, 2018, 11, 8.	0.9	2
33	Whole-exome sequencing reveals doubly novel heterozygous Myosin Binding Protein C and Titin mutations in a Chinese patient with severe dilated cardiomyopathy. Cardiology in the Young, 2018, 28, 1410-1414.	0.8	4
34	The genetic spectrum of familial hypercholesterolemia in the central south region of China. Atherosclerosis, 2017, 258, 84-88.	0.8	22
35	A novel splice-site mutation of WRN (c.IVS28+2T>C) identified in a consanguineous family with Werner Syndrome. Molecular Medicine Reports, 2017, 15, 3735-3738.	2.4	2
36	A mutation of beta-tropomyosin gene in a Chinese family with distal arthrogryposis type I. International Journal of Clinical and Experimental Pathology, 2017, 10, 11137-11142.	0.5	3

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37	Microduplications of 10q24 Detected in Two Chinese Patients with Split-hand/foot Malformation Type 3. Annals of Clinical and Laboratory Science, 2017, 47, 754-757.	0.2	2