Yiping Shen

List of Publications by Year in Descending Order

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Version: 2024-04-28

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

113 papers	4,088 citations	25 h-index	62 g-index
130 ext. papers	4,866 ext. citations	5 avg, IF	4.7 L-index

#	Paper	IF	Citations
113	May Cause Wnt-Fzd Signaling Pathway-Related Nephroblastoma in Children.. *Journal of Biomedical Nanotechnology*, **2022**, 18, 527-534	4	
112	De novo ATP1A2 variants in two Chinese children with alternating hemiplegia of childhood upgraded the gene-disease relationship and variant classification: a case report. *BMC Medical Genomics*, **2021**, 14, 95	3.7	0
111	A novel and recurrent KLHL40 pathogenic variants in a Chinese family of multiple affected neonates with nemaline myopathy 8. *Molecular Genetics & Genomic Medicine*, **2021**, 9, e1683	2.3	1
110	Next generation sequencing in children with unexplained epilepsy: A retrospective cohort study. *Brain and Development*, **2021**, 43, 1004-1012	2.2	
109	A High Proportion of Novel ACAN Mutations and Their Prevalence in a Large Cohort of Chinese Short Stature Children. *Journal of Clinical Endocrinology and Metabolism*, **2021**, 106, e2711-e2719	5.6	8
108	Novel compound heterozygous frameshift variants in WDR81 associated with congenital hydrocephalus 3 with brain anomalies: First Chinese prenatal case confirms WDR81 involvement. *Molecular Genetics & Genomic Medicine*, **2021**, 9, e1624	2.3	1
107	Long-read sequencing identified a novel nonsense and a de novo missense of PPA2 in trans in a Chinese patient with autosomal recessive infantile sudden cardiac failure. *Clinica Chimica Acta*, **2021**, 519, 163-171	6.2	0
106	HPDL deficiency causes a neuromuscular disease by impairing the mitochondrial respiration. *Journal of Genetics and Genomics*, **2021**, 48, 727-736	4	2
105	Trio exome sequencing identified a novel de novo WASF1 missense variant leading to recurrent site substitution in a Chinese patient with developmental delay, microcephaly, and early-onset seizures: A mutational hotspot p.Trp161 and literature review. *Clinica Chimica Acta*, **2021**, 523, 10-18	6.2	1
104	CNV profiles of Chinese pediatric patients with developmental disorders. *Genetics in Medicine*, **2021** , 23, 669-678	8.1	7
103	Antley-Bixler syndrome arising from compound heterozygotes in the P450 oxidoreductase gene: a case report.. *Translational Pediatrics*, **2021**, 10, 3309-3318	4.2	0
102	Whole-exome sequencing identified novel compound heterozygous variants in a Chinese neonate with liver failure and review of literature. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1515	2.3	0
101	A novel pathogenic frameshift variant unmasked by a large de novo deletion at 13q21.33-q31.1 in a Chinese patient with neuronal ceroid lipofuscinosis type 5. *BMC Medical Genetics*, **2020**, 21, 100	2.1	1
100	Novel compound heterozygous pathogenic variants in ASCC1 in a Chinese patient with spinal muscular atrophy with congenital bone fractures 2 : Evidence supporting a "Definitive" gene-disease relationship. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1212	2.3	5
99	A novel variant of IHH in a Chinese family with brachydactyly type 1. *BMC Medical Genetics*, **2020**, 21, 60	2.1	2
98	Applications of cerebrospinal fluid circulating tumor DNA in the diagnosis of gliomas. *Japanese Journal of Clinical Oncology*, **2020**, 50, 325-332	2.8	8
97	Cardio-facio-cutaneous syndrome-associated pathogenic MAP2K1 variants activate autophagy. *Gene*, **2020**, 733, 144369	3.8	4

96	Novel compound heterozygous variant of BSCL2 identified by whole exome sequencing and multiplex ligation-dependent probe amplification in an infant with congenital generalized lipodystrophy. *Molecular Medicine Reports*, **2020**, 21, 2296-2302	2.9	1
95	The phenotypic spectrum of Kabuki syndrome in patients of Chinese descent: A case series. *American Journal of Medical Genetics, Part A*, **2020**, 182, 640-651	2.5	3
94	An Initial Survey of the Performances of Exome Variant Analysis and Clinical Reporting Among Diagnostic Laboratories in China. *Frontiers in Genetics*, **2020**, 11, 582637	4.5	0
93	The first familial NSD2 cases with a novel variant in a Chinese father and daughter with atypical WHS facial features and a 7.5-year follow-up of growth hormone therapy. *BMC Medical Genomics*, **2020**, 13, 181	3.7	2
92	The first two Chinese Myhre syndrome patients with the recurrent SMAD4 pathogenic variants: Functional consequences and clinical diversity. *Clinica Chimica Acta*, **2020**, 500, 128-134	6.2	2
91	Novel genotypes and phenotypes among Chinese patients with Floating-Harbor syndrome. *Orphanet Journal of Rare Diseases*, **2019**, 14, 144	4.2	7
90	Three additional de novo CTCF mutations in Chinese patients help to define an emerging neurodevelopmental disorder. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2019**, 181, 218-225	3.1	5
89	Two Chinese Xia-Gibbs syndrome patients with partial growth hormone deficiency. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e00596	2.3	6
88	Clinical Presentation and Novel Pathogenic Variants among 68 Chinese Neurofibromatosis 1 Children. *Genes*, **2019**, 10,	4.2	5
87	New insights from unbiased panel and whole-exome sequencing in a large Chinese cohort with disorders of sex development. *European Journal of Endocrinology*, **2019**, 181, 311-323	6.5	5
86	Novel Compound Heterozygous Variants in the Gene in a Genetically Male Patient with Female External Genitalia. *JCRPE Journal of Clinical Research in Pediatric Endocrinology*, **2019**, 11, 211-217	1.9	6
85	Biallelic ERBB3 loss-of-function variants are associated with a novel multisystem syndrome without congenital contracture. *Orphanet Journal of Rare Diseases*, **2019**, 14, 265	4.2	3
84	Genome analysis and knowledge-driven variant interpretation with TGex. *BMC Medical Genomics*, **2019**, 12, 200	3.7	12
83	CYP24A1 Variants in Two Chinese Patients with Idiopathic Infantile Hypercalcemia. *Fetal and Pediatric Pathology*, **2019**, 38, 44-56	1.7	1
82	TBX6-associated congenital scoliosis (TACS) as a clinically distinguishable subtype of congenital scoliosis: further evidence supporting the compound inheritance and TBX6 gene dosage model. *Genetics in Medicine*, **2019**, 21, 1548-1558	8.1	36
81	Evaluation of copy number variant detection from panel-based next-generation sequencing data. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e00513	2.3	25
80	Novel pathogenic RECQL4 variants in Chinese patients with Rothmund-Thomson syndrome. *Gene*, **2018**, 654, 110-115	3.8	5
79	A de novo 921 Kb microdeletion at 11q13.1 including neurexin 2 in a boy with developmental delay, deficits in speech and language without autistic behaviors. *European Journal of Medical Genetics*, **2018**, 61, 607-611	2.6	3

78	Three-generation family with novel contiguous gene deletion on chromosome 2p22 associated with thoracic aortic aneurysm syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 560-569	2.5	9
77	Novel compound heterozygous variants in the LHCGR gene identified in a subject with Leydig cell hypoplasia type 1. *Journal of Pediatric Endocrinology and Metabolism*, **2018**, 31, 239-245	1.6	7
76	A rare unbalanced Y:autosome translocation in a Turner syndrome patient. *Journal of Pediatric Endocrinology and Metabolism*, **2018**, 31, 349-353	1.6	2
75	Proband-only medical exome sequencing as a cost-effective first-tier genetic diagnostic test for patients without prior molecular tests and clinical diagnosis in a developing country: the China experience. *Genetics in Medicine*, **2018**, 20, 1045-1053	8.1	38
74	Opposing Tumor-Promoting and -Suppressive Functions of Rictor/mTORC2 Signaling in Adult Glioma and Pediatric SHH Medulloblastoma. *Cell Reports*, **2018**, 24, 463-478.e5	10.6	11
73	Whole-exome sequencing reveals known and novel variants in a cohort of intracranial vertebral-basilar artery dissection (IVAD). *Journal of Human Genetics*, **2018**, 63, 1119-1128	4.3	10
72	A rare exonic NRXN3 deletion segregating with neurodevelopmental and neuropsychiatric conditions in a three-generation Chinese family. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2018**, 177, 589-595	3.5	11
71	Increased transactivation and impaired repression of Ecatenin-mediated transcription associated with a novel SOX3 missense mutation in an X-linked hypopituitarism pedigree with modest growth failure. *Molecular and Cellular Endocrinology*, **2018**, 478, 133-140	4.4	4
70	Clinical and molecular genetic characterization of two patients with mutations in the phosphoglucomutase 1 (PGM1) gene. *Journal of Pediatric Endocrinology and Metabolism*, **2018**, 31, 781-788	1.6	6
69	SOPH Syndrome with Growth Hormone Deficiency, Normal Bone Age, and Novel Compound Heterozygous Mutations in NBAS. *Fetal and Pediatric Pathology*, **2018**, 37, 404-410	1.7	10
68	Trio-R: a script for assessing maternity and paternity in trio studies performed on Agilent chromosomal microarrays. *BMC Medical Informatics and Decision Making*, **2018**, 18, 91	3.6	1
67	Targeted exome sequencing identified a novel mutation hotspot and a deletion in Chinese primary hypertrophic osteoarthropathy patients. *Clinica Chimica Acta*, **2018**, 487, 264-269	6.2	
66	Description of the molecular and phenotypic spectrum of Wiedemann-Steiner syndrome in Chinese patients. *Orphanet Journal of Rare Diseases*, **2018**, 13, 178	4.2	20
65	Approaches to Genetic Testing **2018**, 373-377		
64	Exome Sequencing Identifies De Novo DYNC1H1 Mutations Associated With Distal Spinal Muscular Atrophy and Malformations of Cortical Development. *Journal of Child Neurology*, **2017**, 32, 379-386	2.5	5
63	Novel pathogenic ACAN variants in non-syndromic short stature patients. *Clinica Chimica Acta*, **2017**, 469, 126-129	6.2	28
62	Genetic analysis of Mayer-Rokitansky-Kuster-Hauser syndrome in a large cohort of families. *Fertility and Sterility*, **2017**, 108, 145-151.e2	4.8	21
61	Prenatal and early diagnosis of Chinese 3-M syndrome patients with novel pathogenic variants. *Clinica Chimica Acta*, **2017**, 474, 159-164	6.2	4

60	Biallelic mutations in GPD1 gene in a Chinese boy mainly presented with obesity, insulin resistance, fatty liver, and short stature. *American Journal of Medical Genetics, Part A*, **2017**, 173, 3189-3194	2.5	10
59	Evaluation of three read-depth based CNV detection tools using whole-exome sequencing data. *Molecular Cytogenetics*, **2017**, 10, 30	2	55
58	CNVbase: Batch identification of novel and rare copy number variations based on multi-ethnic population data. *Journal of Genetics and Genomics*, **2017**, 44, 367-370	4	1
57	Further defining the critical genes for the 4q21 microdeletion disorder. *American Journal of Medical Genetics, Part A*, **2017**, 173, 120-125	2.5	6
56	Establishment of Patient-Derived Tumor Xenograft Models of Epithelial Ovarian Cancer for Preclinical Evaluation of Novel Therapeutics. *Clinical Cancer Research*, **2017**, 23, 1263-1273	12.9	67
55	Clinical and Molecular Characterization of Patients with Fructose 1,6-Bisphosphatase Deficiency. *International Journal of Molecular Sciences*, **2017**, 18,	6.3	17
54	Next-generation sequencing analysis of TSHR in 384 Chinese subclinical congenital hypothyroidism (CH) and CH patients. *Clinica Chimica Acta*, **2016**, 462, 127-132	6.2	23
53	A rare occurrence of two large de novo duplications on 1q42-q44 and 9q21.12-q21.33. *Gene*, **2016**, 594, 59-65	3.8	1
52	The presence of two rare genomic syndromes, 1q21 deletion and Xq28 duplication, segregating independently in a family with intellectual disability. *Molecular Cytogenetics*, **2016**, 9, 74	2	5
51	Causal variants screened by whole exome sequencing in a patient with maternal uniparental isodisomy of chromosome 10 and a complicated phenotype. *Experimental and Therapeutic Medicine*, **2016**, 11, 2247-2253	2.1	3
50	A microdeletion at Xq22.2 implicates a glycine receptor GLRA4 involved in intellectual disability, behavioral problems and craniofacial anomalies. *BMC Neurology*, **2016**, 16, 132	3.1	10
49	Copy number variations in 119 Chinese children with idiopathic short stature identified by the custom genome-wide microarray. *Molecular Cytogenetics*, **2016**, 9, 16	2	9
48	Comparative deletion mapping at 1p31.3-p32.2 implies NFIA responsible for intellectual disability coupled with macrocephaly and the presence of several other genes for syndromic intellectual disability. *Molecular Cytogenetics*, **2016**, 9, 24	2	10
47	de novo interstitial deletions at the 11q23.3-q24.2 region. *Molecular Cytogenetics*, **2016**, 9, 39	2	1
46	Thyroglobulin gene mutations in Chinese patients with congenital hypothyroidism. *Molecular and Cellular Endocrinology*, **2016**, 423, 60-6	4.4	18
45	Engineering microdeletions and microduplications by targeting segmental duplications with CRISPR. *Nature Neuroscience*, **2016**, 19, 517-22	25.5	39
44	Genome-wide copy number variant analysis for congenital ventricular septal defects in Chinese Han population. *BMC Medical Genomics*, **2016**, 9, 2	3.7	18
43	A novel de novo microdeletion at 17q11.2 adjacent to NF1 gene associated with developmental delay, short stature, microcephaly and dysmorphic features. *Molecular Cytogenetics*, **2016**, 9, 41	2	7

42	A New Subtype of Multiple Synostoses Syndrome Is Caused by a Mutation in GDF6 That Decreases Its Sensitivity to Noggin and Enhances Its Potency as a BMP Signal. *Journal of Bone and Mineral Research*, **2016**, 31, 882-9	6.3	17
41	Diagnostic value of multiple café-au-lait macules for neurofibromatosis 1 in Chinese children. *Journal of Dermatology*, **2016**, 43, 537-42	1.6	11
40	X-linked adrenal hypoplasia congenita and hypogonadotropic hypogonadism: Identification and in vitro study of a novel small indel in the NR0B1 gene. *Molecular Medicine Reports*, **2016**, 13, 4039-45	2.9	1
39	Novel mutations in the CYP11B2 gene causing aldosterone synthase deficiency. *Molecular Medicine Reports*, **2016**, 13, 3127-32	2.9	7
38	Next-generation sequencing analysis of DUOX2 in 192 Chinese subclinical congenital hypothyroidism (SCH) and CH patients. *Clinica Chimica Acta*, **2016**, 458, 30-4	6.2	34
37	Autistic children exhibit decreased levels of essential Fatty acids in red blood cells. *International Journal of Molecular Sciences*, **2015**, 16, 10061-76	6.3	65
36	EPHA4 haploinsufficiency is responsible for the short stature of a patient with 2q35-q36.2 deletion and Waardenburg syndrome. *BMC Medical Genetics*, **2015**, 16, 23	2.1	5
35	Exome sequencing reveals a novel PTHLH mutation in a Chinese pedigree with brachydactyly type E and short stature. *Clinica Chimica Acta*, **2015**, 446, 9-14	6.2	17
34	PAX8 pathogenic variants in Chinese patients with congenital hypothyroidism. *Clinica Chimica Acta*, **2015**, 450, 322-6	6.2	14
33	A behavioral defect of temporal association memory in mice that partly lack dopamine reuptake transporter. *Scientific Reports*, **2015**, 5, 17461	4.9	7
32	De novo mutations in ARID1B associated with both syndromic and non-syndromic short stature. *BMC Genomics*, **2015**, 16, 701	4.5	17
31	Sequence Variant Interpretation 2.0: Perspective on New Guidelines for Sequence Variant Classification. *Clinical Chemistry*, **2015**, 61, 1317-9	5.5	5
30	TBX6 null variants and a common hypomorphic allele in congenital scoliosis. *New England Journal of Medicine*, **2015**, 372, 341-50	59.2	171
29	Dystrophin is a tumor suppressor in human cancers with myogenic programs. *Nature Genetics*, **2014**, 46, 601-6	36.3	104
28	The Rapidly Emerging Role for Whole Exome Sequencing in Clinical Genetics. *Current Genetic Medicine Reports*, **2014**, 2, 103-112	2.2	2
27	When a "disease-causing mutation" is not a pathogenic variant. *Clinical Chemistry*, **2014**, 60, 711-3	5.5	19
26	Familial 46,XY sex reversal without campomelic dysplasia caused by a deletion upstream of the SOX9 gene. *Molecular and Cellular Endocrinology*, **2014**, 393, 1-7	4.4	19
25	LIN28 is involved in glioma carcinogenesis and predicts outcomes of glioblastoma multiforme patients. *PLoS ONE*, **2014**, 9, e86446	3.7	24

24	Clinical and molecular evaluations of siblings with "pure" 11q23.3-qter trisomy or reciprocal monosomy due to a familial translocation t (10;11) (q26;q23.3). *Molecular Cytogenetics*, **2014**, 7, 101	2	4
23	Whole exome sequencing to identify genetic causes of short stature. *Hormone Research in Paediatrics*, **2014**, 82, 44-52	3.3	57
22	Chromosome microarray testing for patients with congenital heart defects reveals novel disease causing loci and high diagnostic yield. *BMC Genomics*, **2014**, 15, 1127	4.5	60
21	Novel frame-shift mutations of GLI3 gene in non-syndromic postaxial polydactyly patients. *Clinica Chimica Acta*, **2014**, 433, 195-9	6.2	14
20	SOX12 and NRSN2 are candidate genes for 20p13 subtelomeric deletions associated with developmental delay. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2013**, 162B, 832-40	3.5	13
19	Otopalatodigital syndrome type 2 in a male infant: A case report with a novel sequence variation. *Journal of Pediatric Genetics*, **2013**, 2, 33-6	0.7	4
18	Association between MTHFR gene polymorphisms and the risk of autism spectrum disorders: a meta-analysis. *Autism Research*, **2013**, 6, 384-92	5.1	66
17	GAP-43 dependency defines distinct effects of netrin-1 on cortical and spinal neurite outgrowth and directional guidance. *International Journal of Developmental Neuroscience*, **2013**, 31, 11-20	2.7	13
16	Exome and whole-genome sequencing as clinical tests: a transformative practice in molecular diagnostics. *Clinical Chemistry*, **2012**, 58, 1507-9	5.5	24
15	Massive genomic data processing and deep analysis. *Proceedings of the VLDB Endowment*, **2012**, 5, 1906-1909	3.1	5
14	Age- and gender-dependent obesity in individuals with 16p11.2 deletion. *Journal of Genetics and Genomics*, **2011**, 38, 403-9	4	11
13	Genome-wide association of copy-number variation reveals an association between short stature and the presence of low-frequency genomic deletions. *American Journal of Human Genetics*, **2011**, 89, 751-9	11	52
12	Intra-family phenotypic heterogeneity of 16p11.2 deletion carriers in a three-generation Chinese family. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2011**, 156, 225-32	3.5	27
11	Clinical genetic testing for patients with autism spectrum disorders. *Pediatrics*, **2010**, 125, e727-35	7.4	281
10	Cognitive and behavioral characterization of 16p11.2 deletion syndrome. *Journal of Developmental and Behavioral Pediatrics*, **2010**, 31, 649-57	2.4	89
9	Deletions of NRXN1 (neurexin-1) predispose to a wide spectrum of developmental disorders. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2010**, 153B, 937-47	3.5	156
8	Microarray-based genomic DNA profiling technologies in clinical molecular diagnostics. *Clinical Chemistry*, **2009**, 55, 659-69	5.5	47
7	Designing a simple multiplex ligation-dependent probe amplification (MLPA) assay for rapid detection of copy number variants in the genome. *Journal of Genetics and Genomics*, **2009**, 36, 257-65	4	36

6	Disruption of neurexin 1 associated with autism spectrum disorder. *American Journal of Human Genetics*, **2008**, 82, 199-207	11	457
5	Both cell-autonomous and cell non-autonomous functions of GAP-43 are required for normal patterning of the cerebellum in vivo. *Cerebellum*, **2008**, 7, 451-66	4.3	16
4	Association between microdeletion and microduplication at 16p11.2 and autism. *New England Journal of Medicine*, **2008**, 358, 667-75	59.2	1249
3	Development of a focused oligonucleotide-array comparative genomic hybridization chip for clinical diagnosis of genomic imbalance. *Clinical Chemistry*, **2007**, 53, 2051-9	5.5	48
2	Failure to express GAP-43 leads to disruption of a multipotent precursor and inhibits astrocyte differentiation. *Molecular and Cellular Neurosciences*, **2004**, 26, 390-405	4.8	32
1	Growth-associated protein-43 is required for commissural axon guidance in the developing vertebrate nervous system. *Journal of Neuroscience*, **2002**, 22, 239-47	6.6	125