Yiping Shen

List of Publications by Year
in descending order

116 papers	5,407 citations	201674 27 h-index	91884 69 g-index
130 all docs	130 docs citations	130 times ranked	9554 citing authors

#	Article	IF	Citations
1	Genome sequencing demonstrates high diagnostic yield in children with undiagnosed global developmental delay/intellectual disability: A prospective study. Human Mutation, 2022, 43, 568-581.	2.5	12
2	Fos-Related Antigen 1 May Cause Wnt-Fzd Signaling Pathway-Related Nephroblastoma in Children. Journal of Biomedical Nanotechnology, 2022, 18, 527-534.	1.1	0
3	Novel and recurrent ASPM mutations of founder effect in Chinese population. Brain and Development, 2022, 44, 540-545.	1.1	2
4	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.	3.6	22
5	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.	1.2	5
6	De novo ATP1A2 variants in two Chinese children with alternating hemiplegia of childhood upgraded the geneâ€“disease relationship and variant classification: a case report. BMC Medical Genomics, 2021, 14, 95.	1.5	4
7	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.	1.2	4
8	Next generation sequencing in children with unexplained epilepsy: A retrospective cohort study. Brain and Development, 2021, 43, 1004-1012.	1.1	2
9	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.	1.1	3
10	HPDL deficiency causes a neuromuscular disease by impairing the mitochondrial respiration. Journal of Genetics and Genomics, 2021, 48, 727-736.	3.9	5
11	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.	1.1	5
12	CNV profiles of Chinese pediatric patients with developmental disorders. Genetics in Medicine, 2021, 23, 669-678.	2.4	17
13	Novel truncating variant of MN1 penultimate exon identified in a Chinese patient with newly recognized MN1 Câ€terminal truncation syndrome: Case report and literature review. International Journal of Developmental Neuroscience, 2021, , .	1.6	2
14	Antley-Bixler syndrome arising from compound heterozygotes in the P450 oxidoreductase gene: a case report. Translational Pediatrics, 2021, 10, 3309-3318.	1.2	2
15	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.	1.1	5
16	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.	1.2	8
17	An Initial Survey of the Performances of Exome Variant Analysis and Clinical Reporting Among Diagnostic Laboratories in China. Frontiers in Genetics, 2020, 11, 582637.	2.3	2
18	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.	1.5	6

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19	Wholeâ€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.	1.2	2
20	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	3
21	Novel compound heterozygous pathogenic variants in ASCC1 in a Chinese patient with spinal muscular atrophy with congenital bone fractures 2 : Evidence supporting a "Definitive" geneâ€disease relationship. Molecular Genetics & Genomic Medicine, 2020, 8, e1212.	1.2	6
22	A novel variant of IHH in a Chinese family with brachydactyly type 1. BMC Medical Genetics, 2020, 21, 60.	2.1	4
23	Applications of cerebrospinal fluid circulating tumor DNA in the diagnosis of gliomas. Japanese Journal of Clinical Oncology, 2020, 50, 325-332.	1.3	26
24	Cardio-facio-cutaneous syndrome-associated pathogenic MAP2K1 variants activate autophagy. Gene, 2020, 733, 144369.	2.2	14
25	Novel compound heterozygous variant of BSCL2 identified by whole exome sequencing and multiplex ligationâ€dependent probe amplification in an infant with congenital generalized lipodystrophy. Molecular Medicine Reports, 2020, 21, 2296-2302.	2.4	1
26	Clinical Presentation and Novel Pathogenic Variants among 68 Chinese Neurofibromatosis 1 Children. Genes, 2019, 10, 847.	2.4	11
27	Novel genotypes and phenotypes among Chinese patients with Floating-Harbor syndrome. Orphanet Journal of Rare Diseases, 2019, 14, 144.	2.7	10
28	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.	1.6	24
29	Two Chinese Xiaâ€Gibbs syndrome patients with partial growth hormone deficiency. Molecular Genetics & Genomic Medicine, 2019, 7, e00596.	1.2	14
30	Biallelic ERBB3 loss-of-function variants are associated with a novel multisystem syndrome without congenital contracture. Orphanet Journal of Rare Diseases, 2019, 14, 265.	2.7	4
31	Genome analysis and knowledge-driven variant interpretation with TGex. BMC Medical Genomics, 2019, 12, 200.	1.5	30
32	CYP24A1 Variants in Two Chinese Patients with Idiopathic Infantile Hypercalcemia. Fetal and Pediatric Pathology, 2019, 38, 44-56.	0.7	7
33	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.	2.4	60
34	Evaluation of copy number variant detection from panelâ€based nextâ€generation sequencing data. Molecular Genetics & Genomic Medicine, 2019, 7, e00513.	1.2	35
35	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.	3.7	13
36	Novel Compound Heterozygous Variants in the <i>LHCGR</i> Gene in a Genetically Male Patient with Female External Genitalia. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2019, 11, 211-217.	0.9	9

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37	Novel pathogenic RECQL4 variants in Chinese patients with Rothmund-Thomson syndrome. Gene, 2018, 654, 110-115.	2.2	8
38	A de novo 921â€¯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.	1.3	4
39	Threeâ€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.	1.2	14
40	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.	0.9	9
41	A rare unbalanced Y:autosome translocation in a Turner syndrome patient. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 349-353.	0.9	2
42	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.	2.4	64
43	Nextâ€generation sequencing based molecular testing is an equalizer for diagnostic service of rare genetic disorders in China. Pediatric Investigation, 2018, 2, 96-97.	1.4	19
44	SOPH Syndrome with Growth Hormone Deficiency, Normal Bone Age, and Novel Compound Heterozygous Mutations in NBAS. Fetal and Pediatric Pathology, 2018, 37, 404-410.	0.7	11
45	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.	3.0	1
46	Targeted exome sequencing identified a novel mutation hotspot and a deletion in Chinese primary hypertrophic osteoarthropathy patients. Clinica Chimica Acta, 2018, 487, 264-269.	1.1	3
47	Description of the molecular and phenotypic spectrum of Wiedemann-Steiner syndrome in Chinese patients. Orphanet Journal of Rare Diseases, 2018, 13, 178.	2.7	30
48	Opposing Tumor-Promoting and -Suppressive Functions of Rictor/mTORC2 Signaling in Adult Glioma and Pediatric SHH Medulloblastoma. Cell Reports, 2018, 24, 463-478.e5.	6.4	21
49	Whole-exome sequencing reveals known and novel variants in a cohort of intracranial vertebralâ€“basilar artery dissection (IVAD). Journal of Human Genetics, 2018, 63, 1119-1128.	2.3	21
50	A rare exonic NRXN3 deletion segregating with neurodevelopmental and neuropsychiatric conditions in a threeâ€generation Chinese family. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 589-595.	1.7	22
51	Increased transactivation and impaired repression of Î²-catenin-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.	3.2	5
52	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.	0.9	7
53	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.	1.4	10
54	Novel pathogenic ACAN variants in non-syndromic short stature patients. Clinica Chimica Acta, 2017, 469, 126-129.	1.1	35

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55	Genetic analysis of Mayer-Rokitansky-Kuster-Hauser syndrome in a large cohort of families. Fertility and Sterility, 2017, 108, 145-151.e2.	1.0	40
56	Prenatal and early diagnosis of Chinese 3-M syndrome patients with novel pathogenic variants. Clinica Chimica Acta, 2017, 474, 159-164.	1.1	9
57	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.	1.2	19
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.	3.9	1
59	Further defining the critical genes for the 4q21 microdeletion disorder. American Journal of Medical Genetics, Part A, 2017, 173, 120-125.	1.2	17
60	Establishment of Patient-Derived Tumor Xenograft Models of Epithelial Ovarian Cancer for Preclinical Evaluation of Novel Therapeutics. Clinical Cancer Research, 2017, 23, 1263-1273.	7.0	95
61	Clinical and Molecular Characterization of Patients with Fructose 1,6-Bisphosphatase Deficiency. International Journal of Molecular Sciences, 2017, 18, 857.	4.1	27
62	Evaluation of three read-depth based CNV detection tools using whole-exome sequencing data. Molecular Cytogenetics, 2017, 10, 30.	0.9	87
63	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.	0.9	15
64	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, 882-889.	2.8	24
65	Diagnostic value of multiple cafÃ©â€auâ€lait macules for neurofibromatosis 1 in Chinese children. Journal of Dermatology, 2016, 43, 537-542.	1.2	16
66	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, 4039-4045.	2.4	3
67	Novel mutations in the CYP11B2 gene causing aldosterone synthase deficiency. Molecular Medicine Reports, 2016, 13, 3127-3132.	2.4	12
68	Next-generation sequencing analysis of DUOX2 in 192 Chinese subclinical congenital hypothyroidism (SCH) and CH patients. Clinica Chimica Acta, 2016, 458, 30-34.	1.1	46
69	Next-generation sequencing analysis of TSHR in 384 Chinese subclinical congenital hypothyroidism (CH) and CH patients. Clinica Chimica Acta, 2016, 462, 127-132.	1.1	26
70	A rare occurrence of two large de novo duplications on 1q42-q44 and 9q21.12-q21.33. Gene, 2016, 594, 59-65.	2.2	4
71	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.	0.9	7
72	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.	1.8	7

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73	A microdeletion at Xq22.2 implicates a glycine receptor GLRA4 involved in intellectual disability, behavioral problems and craniofacial anomalies. BMC Neurology, 2016, 16, 132.	1.8	21
74	Copy number variations in 119 Chinese children with idiopathic short stature identified by the custom genome-wide microarray. Molecular Cytogenetics, 2016, 9, 16.	0.9	12
75	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.	0.9	17
76	de novo interstitial deletions at the 11q23.3-q24.2 region. Molecular Cytogenetics, 2016, 9, 39.	0.9	2
77	Thyroglobulin gene mutations in Chinese patients with congenital hypothyroidism. Molecular and Cellular Endocrinology, 2016, 423, 60-66.	3.2	23
78	Engineering microdeletions and microduplications by targeting segmental duplications with CRISPR. Nature Neuroscience, 2016, 19, 517-522.	14.8	72
79	Genome-wide copy number variant analysis for congenital ventricular septal defects in Chinese Han population. BMC Medical Genomics, 2015, 9, 2.	1.5	20
80	Otopalatodigital syndrome type 2 in a male infant: A case report with a novel sequence variation. Journal of Pediatric Genetics, 2015, 02, 033-036.	0.7	5
81	A behavioral defect of temporal association memory in mice that partly lack dopamine reuptake transporter. Scientific Reports, 2015, 5, 17461.	3.3	10
82	De novo mutations in ARID1B associated with both syndromic and non-syndromic short stature. BMC Genomics, 2015, 16, 701.	2.8	27
83	Sequence Variant Interpretation 2.0: Perspective on New Guidelines for Sequence Variant Classification. Clinical Chemistry, 2015, 61, 1317-1319.	3.2	5
84	TBX6 Null Variants and a Common Hypomorphic Allele in Congenital Scoliosis. New England Journal of Medicine, 2015, 372, 341-350.	27.0	239
85	Autistic Children Exhibit Decreased Levels of Essential Fatty Acids in Red Blood Cells. International Journal of Molecular Sciences, 2015, 16, 10061-10076.	4.1	81
86	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
87	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.	1.1	20
88	PAX8 pathogenic variants in Chinese patients with congenital hypothyroidism. Clinica Chimica Acta, 2015, 450, 322-326.	1.1	17
89	LIN28 Is Involved in Glioma Carcinogenesis and Predicts Outcomes of Glioblastoma Multiforme Patients. PLoS ONE, 2014, 9, e86446.	2.5	31
90	Clinical and molecular evaluations of siblings with â€œpureâ€•11q23.3-qter trisomy or reciprocal monosomy due to a familial translocation t (10;11) (q26;q23.3). Molecular Cytogenetics, 2014, 7, 101.	0.9	7

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91	Whole Exome Sequencing to Identify Genetic Causes of Short Stature. Hormone Research in Paediatrics, 2014, 82, 44-52.	1.8	76
92	Chromosome microarray testing for patients with congenital heart defects reveals novel disease causing loci and high diagnostic yield. BMC Genomics, 2014, 15, 1127.	2.8	74
93	Novel frame-shift mutations of GLI3 gene in non-syndromic postaxial polydactyly patients. Clinica Chimica Acta, 2014, 433, 195-199.	1.1	16
94	Dystrophin is a tumor suppressor in human cancers with myogenic programs. Nature Genetics, 2014, 46, 601-606.	21.4	142
95	The Rapidly Emerging Role for Whole Exome Sequencing in Clinical Genetics. Current Genetic Medicine Reports, 2014, 2, 103-112.	1.9	5
96	When a "Disease-Causing Mutation" Is Not a Pathogenic Variant. Clinical Chemistry, 2014, 60, 711-713.	3.2	23
97	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.	3.2	22
98	SOX12 and NRSN2 are candidate genes for 20p13 subtelomeric deletions associated with developmental delay. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2013, 162, 832-840.	1.7	15
99	Association between <scp>MTHFR</scp> Gene Polymorphisms and the Risk of Autism Spectrum Disorders: A <scp>M</scp>etaâ€Analysis. Autism Research, 2013, 6, 384-392.	3.8	111
100	GAPâ€43 dependency defines distinct effects of netrinâ€1 on cortical and spinal neurite outgrowth and directional guidance. International Journal of Developmental Neuroscience, 2013, 31, 11-20.	1.6	19
101	Massive genomic data processing and deep analysis. Proceedings of the VLDB Endowment, 2012, 5, 1906-1909.	3.8	5
102	Exome and Whole-Genome Sequencing as Clinical Tests: A Transformative Practice in Molecular Diagnostics. Clinical Chemistry, 2012, 58, 1507-1509.	3.2	28
103	Age- and gender-dependent obesity in individuals with 16p11.2 deletion. Journal of Genetics and Genomics, 2011, 38, 403-409.	3.9	17
104	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, 751-759.	6.2	63
105	Intra-family phenotypic heterogeneity of 16p11.2 deletion carriers in a three-generation Chinese family. , 2011, 156, 225-232.		38
106	Cognitive and Behavioral Characterization of 16p11.2 Deletion Syndrome. Journal of Developmental and Behavioral Pediatrics, 2010, 31, 649-657.	1.1	102
107	Deletions of NRXN1 (neurexinâ€1) predispose to a wide spectrum of developmental disorders. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 937-947.	1.7	217
108	Clinical Genetic Testing for Patients With Autism Spectrum Disorders. Pediatrics, 2010, 125, e727-e735.	2.1	339

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109	Microarray-Based Genomic DNA Profiling Technologies in Clinical Molecular Diagnostics. Clinical Chemistry, 2009, 55, 659-669.	3.2	52
110	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, 257-265.	3.9	40
111	Both Cell-Autonomous and Cell Non-Autonomous Functions of GAP-43 are Required for Normal Patterning of the Cerebellum In Vivo. Cerebellum, 2008, 7, 451-466.	2.5	18
112	Association between Microdeletion and Microduplication at 16p11.2 and Autism. New England Journal of Medicine, 2008, 358, 667-675.	27.0	1,476
113	Disruption of Neurexin 1 Associated with Autism Spectrum Disorder. American Journal of Human Genetics, 2008, 82, 199-207.	6.2	545
114	Development of a Focused Oligonucleotide-Array Comparative Genomic Hybridization Chip for Clinical Diagnosis of Genomic Imbalance. Clinical Chemistry, 2007, 53, 2051-2059.	3.2	51
115	Failure to express GAP-43 leads to disruption of a multipotent precursor and inhibits astrocyte differentiation. Molecular and Cellular Neurosciences, 2004, 26, 390-405.	2.2	37
116	Growth-Associated Protein-43 Is Required for Commissural Axon Guidance in the Developing Vertebrate Nervous System. Journal of Neuroscience, 2002, 22, 239-247.	3.6	137