Xianda Wei

List of Publications by Year in descending order

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100 papers	1,385 citations	471509 17 h-index	434195 31 g-index
101 all docs	101 docs citations	101 times ranked	2145 citing authors

#	Article	IF	Citations
1	Clinical utility of noninvasive prenatal screening for expanded chromosome disease syndromes. Genetics in Medicine, 2019, 21, 1998-2006.	2.4	158
2	Copy Number Variation Sequencing for Comprehensive Diagnosis of Chromosome Disease Syndromes. Journal of Molecular Diagnostics, 2014, 16, 519-526.	2.8	106
3	Noninvasive Prenatal Testing for Wilson Disease by Use of Circulating Single-Molecule Amplification and Resequencing Technology (cSMART). Clinical Chemistry, 2015, 61, 172-181.	3.2	85
4	A More Universal Approach to Comprehensive Analysis of Thalassemia Alleles (CATSA). Journal of Molecular Diagnostics, 2021, 23, 1195-1204.	2.8	55
5	Seamless Genetic Conversion of SMN2 to SMN1 via CRISPR/Cpf1 and Single-Stranded Oligodeoxynucleotides in Spinal Muscular Atrophy Patient-Specific Induced Pluripotent Stem Cells. Human Gene Therapy, 2018, 29, 1252-1263.	2.7	50
6	DMD mutation spectrum analysis in 613 Chinese patients with dystrophinopathy. Journal of Human Genetics, 2015, 60, 435-442.	2.3	47
7	TALE nickase mediates high efficient targeted transgene integration at the human multi-copy ribosomal DNA locus. Biochemical and Biophysical Research Communications, 2014, 446, 261-266.	2.1	45
8	In situ genetic correction of F8 intron 22 inversion in hemophilia A patient-specific iPSCs. Scientific Reports, 2016, 6, 18865.	3.3	43
9	Molecular analysis of hearing loss associated with enlarged vestibular aqueduct in the mainland Chinese: a unique SLC26A4 mutation spectrum. Journal of Human Genetics, 2007, 52, 492-497.	2.3	39
10	A ZRS duplication causes syndactyly type IV with tibial hypoplasia. American Journal of Medical Genetics, Part A, 2009, 149A, 816-818.	1.2	35
11	A novel de novo POGZ mutation in a patient with intellectual disability. Journal of Human Genetics, 2016, 61, 357-359.	2.3	31
12	Long-read DNA sequencing fully characterized chromothripsis in a patient with Langerâ€“Giedion syndrome and Cornelia de Lange syndrome-4. Journal of Human Genetics, 2020, 65, 667-674.	2.3	24
13	ssODN-Mediated In-Frame Deletion with CRISPR/Cas9 Restores FVIII Function in Hemophilia A-Patient-Derived iPSCs and ECs. Molecular Therapy - Nucleic Acids, 2019, 17, 198-209.	5.1	23
14	Comprehensive Analysis of Congenital Adrenal Hyperplasia Using Long-Read Sequencing. Clinical Chemistry, 2022, 68, 927-939.	3.2	23
15	Targeted next-generation sequencing identifies novel compound heterozygous mutations of DYNC2H1 in a fetus with short rib-polydactyly syndrome, type III. Clinica Chimica Acta, 2015, 447, 47-51.	1.1	20
16	Enhanced tumor growth inhibition by mesenchymal stem cells derived from iPSCs with targeted integration of interleukin24 into rDNA loci. Oncotarget, 2017, 8, 40791-40803.	1.8	20
17	Eight-Shaped Hatching Increases the Risk of Inner Cell Mass Splitting in Extended Mouse Embryo Culture. PLoS ONE, 2015, 10, e0145172.	2.5	19
18	De novo exonic deletion of KDM6A in a Chinese girl with Kabuki syndrome: A case report and brief literature review. American Journal of Medical Genetics, Part A, 2016, 170, 1613-1621.	1.2	19

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19	A clear bias in parental origin of de novo pathogenic CNVs related to intellectual disability, developmental delay and multiple congenital anomalies. Scientific Reports, 2017, 7, 44446.	3.3	19
20	Paired CRISPR/Cas9 Nickases Mediate Efficient Site-Specific Integration of F9 into rDNA Locus of Mouse ESCs. International Journal of Molecular Sciences, 2018, 19, 3035.	4.1	19
21	De Novo Mutations of CCNK Cause a Syndromic Neurodevelopmental Disorder with Distinctive Facial Dysmorphism. American Journal of Human Genetics, 2018, 103, 448-455.	6.2	17
22	Constructing a database for the relations between CNV and human genetic diseases via systematic text mining. BMC Bioinformatics, 2018, 19, 528.	2.6	16
23	Site-Specific Integration of TRAIL in iPSC-Derived Mesenchymal Stem Cells for Targeted Cancer Therapy. Stem Cells Translational Medicine, 2022, 11, 297-309.	3.3	16
24	Mesenchymal stem cells derived from iPSCs expressing interleukin-24 inhibit the growth of melanoma in the tumor-bearing mouse model. Cancer Cell International, 2020, 20, 33.	4.1	14
25	Mutation analysis of the TUBB8 gene in primary infertile women with oocyte maturation arrest. Journal of Ovarian Research, 2022, 15, 38.	3.0	14
26	Molecular diagnosis for 55 fetuses with skeletal dysplasias by wholeâ€exome sequencing: A retrospective cohort study. Clinical Genetics, 2021, 100, 219-226.	2.0	13
27	Mutational Analyses of the FMR1 Gene in Chinese Pediatric Population of Fragile X Suspects. Journal of Child Neurology, 2015, 30, 803-806.	1.4	12
28	Novel GATAD2B loss-of-function mutations cause intellectual disability in two unrelated cases. Journal of Human Genetics, 2017, 62, 513-516.	2.3	12
29	Translocation breakpoint disrupting the host SNHG14 gene but not coding genes or snoRNAs in typical Prader-Willi syndrome. Journal of Human Genetics, 2019, 64, 647-652.	2.3	12
30	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
31	Cryptic FMR1 mosaic deletion in a phenotypically normal mother of a boy with fragile X syndrome: case report. BMC Medical Genetics, 2014, 15, 125.	2.1	11
32	Novel and reported APC germline mutations in Chinese patients with familial adenomatous polyposis. Gene, 2016, 577, 187-192.	2.2	11
33	Clinical and molecular investigation in Chinese patients with glutaric aciduria type I. Clinica Chimica Acta, 2016, 453, 75-79.	1.1	11
34	Truncating mutations of HIBCH tend to cause severe phenotypes in cases with HIBCH deficiency: a case report and brief literature review. Journal of Human Genetics, 2018, 63, 851-855.	2.3	11
35	Silica nanoparticle is a possible safe carrier for gene therapy. Science Bulletin, 2005, 50, 2323-2327.	1.7	10
36	A New Next-Generation Sequencing-Based Assay for Concurrent Preimplantation Genetic Diagnosis of Charcot-Marie-Tooth Disease Type 1A and Aneuploidy Screening. Journal of Genetics and Genomics, 2016, 43, 155-159.	3.9	10

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37	WDR73 missense mutation causes infantile onset intellectual disability and cerebellar hypoplasia in a consanguineous family. Clinica Chimica Acta, 2017, 464, 24-29.	1.1	10
38	CRISPR/Cas12a-Based Ultrasensitive and Rapid Detection of JAK2 V617F Somatic Mutation in Myeloproliferative Neoplasms. Biosensors, 2021, 11, 247.	4.7	10
39	Pachygyria, seizures, hypotonia, and growth retardation in a patient with an atypical 1.33Mb inherited microduplication at 22q11.23. Gene, 2015, 569, 46-50.	2.2	9
40	Diagnosis of Joubert Syndrome 10 in a Fetus with Suspected Dandy-Walker Variant by WES: A Novel Splicing Mutation in OFD1. BioMed Research International, 2018, 2018, 1-7.	1.9	9
41	Three Novel Mutations in <i>FBN1</i> and <i>TGFBR2</i> in Patients with the Syndromic Form of Thoracic Aortic Aneurysms and Dissections. International Heart Journal, 2018, 59, 1059-1068.	1.0	9
42	Identification of a novel gross deletion of TCOF1 in a Chinese prenatal case with Treacher Collins syndrome. Molecular Genetics & Genomic Medicine, 2020, 8, e1313.	1.2	9
43	Cas12a and Lateral Flow Strip-Based Test for Rapid and Ultrasensitive Detection of Spinal Muscular Atrophy. Biosensors, 2021, 11, 154.	4.7	9
44	Fertilization and neonatal outcomes after early rescue intracytoplasmic sperm injection: a retrospective analysis of 16,769 patients. Archives of Gynecology and Obstetrics, 2022, 306, 249-258.	1.7	9
45	Three novel mutations of STK11 gene in Chinese patients with Peutzâ€“Jeghers syndrome. BMC Medical Genetics, 2016, 17, 77.	2.1	8
46	Next generation sequencing identified two novel mutations in NIPBL and a frame shift mutation in CREBBP in three Chinese children. Orphanet Journal of Rare Diseases, 2019, 14, 45.	2.7	8
47	Increase in diagnostic yield achieved for 174 whole-exome sequencing cases reanalyzed 1â€“2Âyears after initial analysis. Clinica Chimica Acta, 2021, 523, 163-168.	1.1	8
48	Deficiency in GnRH receptor trafficking due to a novel homozygous mutation causes idiopathic hypogonadotropic hypogonadism in three prepubertal siblings. Gene, 2018, 669, 42-46.	2.2	7
49	Development and validation of a haplotypeâ€free technique for nonâ€invasive prenatal diagnosis of spinal muscular atrophy. Journal of Clinical Laboratory Analysis, 2020, 34, e23046.	2.1	7
50	Predominant cellular mitochondrial dysfunction in the TOP3A gene-caused Bloom syndrome-like disorder. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2021, 1867, 166106.	3.8	7
51	Gene Therapy for Hemophilia A: Where We Stand. Current Gene Therapy, 2020, 20, 142-151.	2.0	7
52	Identification of a novel MIP frameshift mutation associated with congenital cataract in a Chinese family by whole-exome sequencing and functional analysis. Eye, 2018, 32, 1359-1364.	2.1	6
53	Restoration of SMN expression in mesenchymal stem cells derived from gene-targeted patient-specific iPSCs. Journal of Molecular Histology, 2018, 49, 27-37.	2.2	6
54	Six novel Mutation analysis of the androgen receptor gene in 17 Chinese patients with androgen insensitivity syndrome. Clinica Chimica Acta, 2020, 506, 180-186.	1.1	6

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55	Loss of PIGK function causes severe infantile encephalopathy and extensive neuronal apoptosis. Human Genetics, 2021, 140, 791-803.	3.8	6
56	Restoration of FVIII Function and Phenotypic Rescue in Hemophilia A Mice by Transplantation of MSCs Derived From F8-Modified iPSCs. Frontiers in Cell and Developmental Biology, 2021, 9, 630353.	3.7	6
57	Noninvasive Prenatal Testing of Methylmalonic Acidemia cblC Type Using the cSMART Assay for MMACHC Gene Mutations. Frontiers in Genetics, 2021, 12, 750719.	2.3	6
58	Rare intracranial cholesterol deposition and a homozygous mutation of LDLR in a familial hypercholesterolemia patient. Gene, 2015, 569, 313-317.	2.2	5
59	PMP22-Related neuropathies and other clinical manifestations in Chinese han patients with charcot-marie-tooth disease type 1. Muscle and Nerve, 2015, 52, 69-75.	2.2	5
60	Two novel NIPBL gene mutations in Chinese patients with Cornelia de Lange syndrome. Gene, 2015, 555, 476-480.	2.2	5
61	Notable Carrier Risks for Individuals Having Two Copies of SMN1 in Spinal Muscular Atrophy Families with 2â€copy Alleles: Estimation Based on Chinese Metaâ€analysis Data. Journal of Genetic Counseling, 2017, 26, 72-78.	1.6	5
62	Gene Therapy for Hemophilia and Duchenne Muscular Dystrophy in China. Human Gene Therapy, 2018, 29, 146-150.	2.7	5
63	Molecular genetic study of 59 Chinese Oculocutaneous albinism families. European Journal of Medical Genetics, 2019, 62, 103709.	1.3	5
64	Detection of Spinal Muscular Atrophy Using a Duplexed Real-Time PCR Approach With Locked Nucleic Acid-Modified Primers. Annals of Laboratory Medicine, 2021, 41, 101-107.	2.5	5
65	Targeted addition of mini-dystrophin into rDNA locus of Duchenne muscular dystrophy patient-derived iPSCs. Biochemical and Biophysical Research Communications, 2021, 545, 40-45.	2.1	5
66	Identification of Five Novel Mutations Causing Rare Lysosomal Storage Diseases. Medical Science Monitor, 2019, 25, 7634-7644.	1.1	5
67	Expression of reconstructive hFVIII in the hrDNA by using hrDNA targeting vector. Science Bulletin, 2005, 50, 2187-2192.	1.7	4
68	A family with partial duplication/deletion 4p due to a balanced t (4; 15) (p16.2; p11.2) translocation. American Journal of Medical Genetics, Part A, 2011, 155, 656-659.	1.2	4
69	An Xp21.3p11.4 duplication observed in a boy with intellectual deficiency and speech delay and his asymptomatic mother. Birth Defects Research Part A: Clinical and Molecular Teratology, 2013, 97, 467-470.	1.6	4
70	De Novo ring chromosome 11 and non-reciprocal translocation of 11p15.3-pter to 21qter in a patient with congenital heart disease. Molecular Cytogenetics, 2015, 8, 88.	0.9	4
71	Novel de novo nonsense mutation of the PHEX gene (p.Lys50Ter) in a Chinese patient with hypophosphatemic rickets. Gene, 2015, 565, 150-154.	2.2	4
72	Partial trisomy 2q33.3-q37.3 in a patient with an inverted duplicated neocentric marker chromosome. Molecular Cytogenetics, 2015, 8, 10.	0.9	4

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73	A novel MSX1 intronic mutation associated with autosomal dominant non-syndromic oligodontia in a large Chinese family pedigree. Clinica Chimica Acta, 2016, 461, 135-140.	1.1	4
74	Targeted exome sequencing identifies novel compound heterozygous mutations in P3H1 in a fetus with osteogenesis imperfecta type VIII. Clinica Chimica Acta, 2017, 464, 170-175.	1.1	4
75	Identification of six novel mutations in five infants with suspected maple syrup urine disease based on blood and urine metabolism screening. Gene, 2019, 710, 9-16.	2.2	4
76	Novel variants in the KERA gene cause autosomal recessive cornea plana in a Chinese family: A case report. Molecular Medicine Reports, 2019, 19, 4711-4718.	2.4	4
77	28 novel mutations identified from 33 Chinese patients with cilia-related kidney disorders. Clinica Chimica Acta, 2020, 501, 207-215.	1.1	4
78	The rare Alus element-mediated chimerism of multiple de novo complex rearrangement sequences in GAN result in giant axonal neuropathy. Clinica Chimica Acta, 2020, 502, 91-98.	1.1	4
79	A novel GJB1 mutation associated with Xâ€linked Charcotâ€“Marieâ€“Tooth disease in a large Chinese family pedigree. Molecular Genetics & Genomic Medicine, 2020, 8, e1127.	1.2	4
80	IL-24 armored CAR19-T cells show enhanced antitumor activity and persistence. Signal Transduction and Targeted Therapy, 2021, 6, 14.	17.1	4
81	An Episomal CRISPR/Cas12a System for Mediating Efficient Gene Editing. Life, 2021, 11, 1262.	2.4	4
82	Ectopic Expression of FVIII in HPCs and MSCs Derived from hiPSCs with Site-Specific Integration of ITGA2B Promoter-Driven BDDF8 Gene in Hemophilia A. International Journal of Molecular Sciences, 2022, 23, 623.	4.1	4
83	A lesson from a reported pathogenic variant in Peutz-Jeghers syndrome: a case report. Familial Cancer, 2017, 16, 417-422.	1.9	3
84	REDBot: Natural language process methods for clinical copy number variation reporting in prenatal and products of conception diagnosis. Molecular Genetics & Genomic Medicine, 2020, 8, e1488.	1.2	3
85	Behavioral and Gene Expression Analysis of Stxbp6-Knockout Mice. Brain Sciences, 2021, 11, 436.	2.3	3
86	Novel variants in OSGEP leading to Galloway-Mowat syndrome by altering its subcellular localization. Clinica Chimica Acta, 2021, 523, 297-303.	1.1	3
87	Identification of four novel mutations in BTK from six Chinese families with X-linked agammaglobulinemia. Clinica Chimica Acta, 2022, 531, 48-55.	1.1	3
88	Cas14a1-Mediated Nucleic Acid Diagnostics for Spinal Muscular Atrophy. Biosensors, 2022, 12, 268.	4.7	3
89	Preâ€•and postnatal overgrowth in a patient with proximal 4p deletion. American Journal of Medical Genetics, Part A, 2008, 146A, 791-794.	1.2	2
90	Generation of reporter hESCs by targeting EGFP at the CD144 locus to facilitate the endothelial differentiation. Development Growth and Differentiation, 2018, 60, 205-215.	1.5	2

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91	Ectopic expression of factor VIII in MSCs and hepatocytes derived from rDNA targeted hESCs. Clinica Chimica Acta, 2019, 495, 656-663.	1.1	2
92	Identification of pathogenic mutations in 6 Chinese families with multiple exostoses by whole-exome sequencing and multiplex ligation-dependent probe amplification. Medicine (United States), 2019, 98, e15692.	1.0	2
93	Prenatal Diagnosis in a Fetus With X-Linked Recessive Chondrodysplasia Punctata: Identification and Functional Study of a Novel Missense Mutation in ARSE. Frontiers in Genetics, 2021, 12, 722694.	2.3	2
94	Investigation of hrDNA targeting vector-mediated tumor-specific suicide gene therapy for hepatocellular carcinoma. Science Bulletin, 2006, 51, 2342-2350.	1.7	1
95	A novel FOXL2 mutation in a Chinese family with blepharophimosis, ptosis, epicanthus inversus syndrome. Human Genome Variation, 2015, 2, 15008.	0.7	1
96	Simultaneous Identification of Both MFSD8 and RDH12 Pathogenic Variants in a Chinese Family Affected With Retinitis Pigmentosa. Frontiers in Genetics, 2021, 12, 715100.	2.3	1
97	A patient with apparently reciprocal translocation and cryptic 10p deletion. American Journal of Medical Genetics, Part A, 2011, 155, 1753-1755.	1.2	0
98	Morphometric analysis and developmental comparison of embryos from carriers with balanced chromosomal rearrangements in preimplantation genetic diagnosis cycles. Reproduction, Fertility and Development, 2016, 28, 1953.	0.4	0
99	NovelGZF1pathogenic variants identified in two Chinese patients with Larsen syndrome. Clinical Genetics, 2021, 99, 281-285.	2.0	0
100	Biochemical and genetic characteristics of 40 neonates with carnitine deficiency. Journal of Central South University (Medical Sciences), 2020, 45, 1164-1171.	0.1	0