

Xianda Wei

List of Publications by Year in descending order

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Version: 2024-02-01

100
papers

1,385
citations

471061

17
h-index

433756

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. <i>Genetics in Medicine</i> , 2019, 21, 1998-2006.	1.1	158
2	Copy Number Variation Sequencing for Comprehensive Diagnosis of Chromosome Disease Syndromes. <i>Journal of Molecular Diagnostics</i> , 2014, 16, 519-526.	1.2	106
3	Noninvasive Prenatal Testing for Wilson Disease by Use of Circulating Single-Molecule Amplification and Resequencing Technology (cSMART). <i>Clinical Chemistry</i> , 2015, 61, 172-181.	1.5	85
4	A More Universal Approach to Comprehensive Analysis of Thalassemia Alleles (CATSA). <i>Journal of Molecular Diagnostics</i> , 2021, 23, 1195-1204.	1.2	55
5	Seamless Genetic Conversion of <i>SMN2</i> to <i>SMN1</i> via CRISPR/Cpf1 and Single-Stranded Oligodeoxynucleotides in Spinal Muscular Atrophy Patient-Specific Induced Pluripotent Stem Cells. <i>Human Gene Therapy</i> , 2018, 29, 1252-1263.	1.4	50
6	DMD mutation spectrum analysis in 613 Chinese patients with dystrophinopathy. <i>Journal of Human Genetics</i> , 2015, 60, 435-442.	1.1	47
7	TALE nickase mediates high efficient targeted transgene integration at the human multi-copy ribosomal DNA locus. <i>Biochemical and Biophysical Research Communications</i> , 2014, 446, 261-266.	1.0	45
8	In situ genetic correction of F8 intron 22 inversion in hemophilia A patient-specific iPSCs. <i>Scientific Reports</i> , 2016, 6, 18865.	1.6	43
9	Molecular analysis of hearing loss associated with enlarged vestibular aqueduct in the mainland Chinese: a unique SLC26A4 mutation spectrum. <i>Journal of Human Genetics</i> , 2007, 52, 492-497.	1.1	39
10	A <i>ZRS</i> duplication causes syndactyly type IV with tibial hypoplasia. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 816-818.	0.7	35
11	A novel de novo POGZ mutation in a patient with intellectual disability. <i>Journal of Human Genetics</i> , 2016, 61, 357-359.	1.1	31
12	Long-read DNA sequencing fully characterized chromothripsis in a patient with Langer-Giedion syndrome and Cornelia de Lange syndrome-4. <i>Journal of Human Genetics</i> , 2020, 65, 667-674.	1.1	24
13	ssODN-Mediated In-Frame Deletion with CRISPR/Cas9 Restores FVIII Function in Hemophilia A-Patient-Derived iPSCs and ECs. <i>Molecular Therapy - Nucleic Acids</i> , 2019, 17, 198-209.	2.3	23
14	Comprehensive Analysis of Congenital Adrenal Hyperplasia Using Long-Read Sequencing. <i>Clinical Chemistry</i> , 2022, 68, 927-939.	1.5	23
15	Targeted next-generation sequencing identifies novel compound heterozygous mutations of DYNC2H1 in a fetus with short rib-polydactyly syndrome, type III. <i>Clinica Chimica Acta</i> , 2015, 447, 47-51.	0.5	20
16	Enhanced tumor growth inhibition by mesenchymal stem cells derived from iPSCs with targeted integration of interleukin24 into rDNA loci. <i>Oncotarget</i> , 2017, 8, 40791-40803.	0.8	20
17	Eight-Shaped Hatching Increases the Risk of Inner Cell Mass Splitting in Extended Mouse Embryo Culture. <i>PLoS ONE</i> , 2015, 10, e0145172.	1.1	19
18	De novo exonic deletion of <i>KDM6A</i> in a Chinese girl with Kabuki syndrome: A case report and brief literature review. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1613-1621.	0.7	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. <i>Scientific Reports</i> , 2017, 7, 44446.	1.6	19
20	Paired CRISPR/Cas9 Nickases Mediate Efficient Site-Specific Integration of F9 into rDNA Locus of Mouse ESCs. <i>International Journal of Molecular Sciences</i> , 2018, 19, 3035.	1.8	19
21	De Novo Mutations of CCNK Cause a Syndromic Neurodevelopmental Disorder with Distinctive Facial Dysmorphism. <i>American Journal of Human Genetics</i> , 2018, 103, 448-455.	2.6	17
22	Constructing a database for the relations between CNV and human genetic diseases via systematic text mining. <i>BMC Bioinformatics</i> , 2018, 19, 528.	1.2	16
23	Site-Specific Integration of <i>TRAIL</i> in iPSC-Derived Mesenchymal Stem Cells for Targeted Cancer Therapy. <i>Stem Cells Translational Medicine</i> , 2022, 11, 297-309.	1.6	16
24	Mesenchymal stem cells derived from iPSCs expressing interleukin-24 inhibit the growth of melanoma in the tumor-bearing mouse model. <i>Cancer Cell International</i> , 2020, 20, 33.	1.8	14
25	Mutation analysis of the TUBB8 gene in primary infertile women with oocyte maturation arrest. <i>Journal of Ovarian Research</i> , 2022, 15, 38.	1.3	14
26	Molecular diagnosis for 55 fetuses with skeletal dysplasias by whole-exome sequencing: A retrospective cohort study. <i>Clinical Genetics</i> , 2021, 100, 219-226.	1.0	13
27	Mutational Analyses of the <i>FMR1</i> Gene in Chinese Pediatric Population of Fragile X Suspects. <i>Journal of Child Neurology</i> , 2015, 30, 803-806.	0.7	12
28	Novel GATAD2B loss-of-function mutations cause intellectual disability in two unrelated cases. <i>Journal of Human Genetics</i> , 2017, 62, 513-516.	1.1	12
29	Translocation breakpoint disrupting the host SNHG14 gene but not coding genes or snRNAs in typical Prader-Willi syndrome. <i>Journal of Human Genetics</i> , 2019, 64, 647-652.	1.1	12
30	Genome sequencing demonstrates high diagnostic yield in children with undiagnosed global developmental delay/intellectual disability: A prospective study. <i>Human Mutation</i> , 2022, 43, 568-581.	1.1	12
31	Cryptic FMR1 mosaic deletion in a phenotypically normal mother of a boy with fragile X syndrome: case report. <i>BMC Medical Genetics</i> , 2014, 15, 125.	2.1	11
32	Novel and reported APC germline mutations in Chinese patients with familial adenomatous polyposis. <i>Gene</i> , 2016, 577, 187-192.	1.0	11
33	Clinical and molecular investigation in Chinese patients with glutaric aciduria type I. <i>Clinica Chimica Acta</i> , 2016, 453, 75-79.	0.5	11
34	Truncating mutations of HIBCH tend to cause severe phenotypes in cases with HIBCH deficiency: a case report and brief literature review. <i>Journal of Human Genetics</i> , 2018, 63, 851-855.	1.1	11
35	Silica nanoparticle is a possible safe carrier for gene therapy. <i>Science Bulletin</i> , 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. <i>Journal of Genetics and Genomics</i> , 2016, 43, 155-159.	1.7	10

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37	WDR73 missense mutation causes infantile onset intellectual disability and cerebellar hypoplasia in a consanguineous family. <i>Clinica Chimica Acta</i> , 2017, 464, 24-29.	0.5	10
38	CRISPR/Cas12a-Based Ultrasensitive and Rapid Detection of JAK2 V617F Somatic Mutation in Myeloproliferative Neoplasms. <i>Biosensors</i> , 2021, 11, 247.	2.3	10
39	Pachygyria, seizures, hypotonia, and growth retardation in a patient with an atypical 1.33Mb inherited microduplication at 22q11.23. <i>Gene</i> , 2015, 569, 46-50.	1.0	9
40	Diagnosis of Joubert Syndrome 10 in a Fetus with Suspected Dandy-Walker Variant by WES: A Novel Splicing Mutation in <i>OFD1</i> . <i>BioMed Research International</i> , 2018, 2018, 1-7.	0.9	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. <i>International Heart Journal</i> , 2018, 59, 1059-1068.	0.5	9
42	Identification of a novel gross deletion of <i>TCOF1</i> in a Chinese prenatal case with Treacher Collins syndrome. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1313.	0.6	9
43	Cas12a and Lateral Flow Strip-Based Test for Rapid and Ultrasensitive Detection of Spinal Muscular Atrophy. <i>Biosensors</i> , 2021, 11, 154.	2.3	9
44	Fertilization and neonatal outcomes after early rescue intracytoplasmic sperm injection: a retrospective analysis of 16,769 patients. <i>Archives of Gynecology and Obstetrics</i> , 2022, 306, 249-258.	0.8	9
45	Three novel mutations of <i>STK11</i> gene in Chinese patients with Peutz-Jeghers syndrome. <i>BMC Medical Genetics</i> , 2016, 17, 77.	2.1	8
46	Next generation sequencing identified two novel mutations in <i>NIPBL</i> and a frame shift mutation in <i>CREBBP</i> in three Chinese children. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 45.	1.2	8
47	Increase in diagnostic yield achieved for 174 whole-exome sequencing cases reanalyzed 2 years after initial analysis. <i>Clinica Chimica Acta</i> , 2021, 523, 163-168.	0.5	8
48	Deficiency in GnRH receptor trafficking due to a novel homozygous mutation causes idiopathic hypogonadotropic hypogonadism in three prepubertal siblings. <i>Gene</i> , 2018, 669, 42-46.	1.0	7
49	Development and validation of a haplotype-free technique for non-invasive prenatal diagnosis of spinal muscular atrophy. <i>Journal of Clinical Laboratory Analysis</i> , 2020, 34, e23046.	0.9	7
50	Predominant cellular mitochondrial dysfunction in the <i>TOP3A</i> gene-caused Bloom syndrome-like disorder. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2021, 1867, 166106.	1.8	7
51	Gene Therapy for Hemophilia A: Where We Stand. <i>Current Gene Therapy</i> , 2020, 20, 142-151.	0.9	7
52	Identification of a novel MIP frameshift mutation associated with congenital cataract in a Chinese family by whole-exome sequencing and functional analysis. <i>Eye</i> , 2018, 32, 1359-1364.	1.1	6
53	Restoration of <i>SMN</i> expression in mesenchymal stem cells derived from gene-targeted patient-specific iPSCs. <i>Journal of Molecular Histology</i> , 2018, 49, 27-37.	1.0	6
54	Six novel Mutation analysis of the androgen receptor gene in 17 Chinese patients with androgen insensitivity syndrome. <i>Clinica Chimica Acta</i> , 2020, 506, 180-186.	0.5	6

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55	Loss of PIGK function causes severe infantile encephalopathy and extensive neuronal apoptosis. <i>Human Genetics</i> , 2021, 140, 791-803.	1.8	6
56	Restoration of FVIII Function and Phenotypic Rescue in Hemophilia A Mice by Transplantation of MSCs Derived From F8-Modified iPSCs. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 630353.	1.8	6
57	Noninvasive Prenatal Testing of Methylmalonic Acidemia cblC Type Using the cSMART Assay for MMAHC Gene Mutations. <i>Frontiers in Genetics</i> , 2021, 12, 750719.	1.1	6
58	Rare intracranial cholesterol deposition and a homozygous mutation of LDLR in a familial hypercholesterolemia patient. <i>Gene</i> , 2015, 569, 313-317.	1.0	5
59	PMP22-Related neuropathies and other clinical manifestations in Chinese han patients with charcot-marie-tooth disease type 1. <i>Muscle and Nerve</i> , 2015, 52, 69-75.	1.0	5
60	Two novel NIPBL gene mutations in Chinese patients with Cornelia de Lange syndrome. <i>Gene</i> , 2015, 555, 476-480.	1.0	5
61	Notable Carrier Risks for Individuals Having Two Copies of <i>SMN1</i> in Spinal Muscular Atrophy Families with 2-copy Alleles: Estimation Based on Chinese Meta-analysis Data. <i>Journal of Genetic Counseling</i> , 2017, 26, 72-78.	0.9	5
62	Gene Therapy for Hemophilia and Duchenne Muscular Dystrophy in China. <i>Human Gene Therapy</i> , 2018, 29, 146-150.	1.4	5
63	Molecular genetic study of 59 Chinese Oculocutaneous albinism families. <i>European Journal of Medical Genetics</i> , 2019, 62, 103709.	0.7	5
64	Detection of Spinal Muscular Atrophy Using a Duplexed Real-Time PCR Approach With Locked Nucleic Acid-Modified Primers. <i>Annals of Laboratory Medicine</i> , 2021, 41, 101-107.	1.2	5
65	Targeted addition of mini-dystrophin into rDNA locus of Duchenne muscular dystrophy patient-derived iPSCs. <i>Biochemical and Biophysical Research Communications</i> , 2021, 545, 40-45.	1.0	5
66	Identification of Five Novel Mutations Causing Rare Lysosomal Storage Diseases. <i>Medical Science Monitor</i> , 2019, 25, 7634-7644.	0.5	5
67	Expression of reconstructive hFVIII in the hrDNA by using hrDNA targeting vector. <i>Science Bulletin</i> , 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. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 656-659.	0.7	4
69	An Xp21.3p11.4 duplication observed in a boy with intellectual deficiency and speech delay and his asymptomatic mother. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 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. <i>Molecular Cytogenetics</i> , 2015, 8, 88.	0.4	4
71	Novel de novo nonsense mutation of the PHEX gene (p.Lys50Ter) in a Chinese patient with hypophosphatemic rickets. <i>Gene</i> , 2015, 565, 150-154.	1.0	4
72	Partial trisomy 2q33.3-q37.3 in a patient with an inverted duplicated neocentric marker chromosome. <i>Molecular Cytogenetics</i> , 2015, 8, 10.	0.4	4

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73	A novel MSX1 intronic mutation associated with autosomal dominant non-syndromic oligodontia in a large Chinese family pedigree. <i>Clinica Chimica Acta</i> , 2016, 461, 135-140.	0.5	4
74	Targeted exome sequencing identifies novel compound heterozygous mutations in P3H1 in a fetus with osteogenesis imperfecta type VIII. <i>Clinica Chimica Acta</i> , 2017, 464, 170-175.	0.5	4
75	Identification of six novel mutations in five infants with suspected maple syrup urine disease based on blood and urine metabolism screening. <i>Gene</i> , 2019, 710, 9-16.	1.0	4
76	Novel variants in the KERA gene cause autosomal recessive cornea plana in a Chinese family: A case report. <i>Molecular Medicine Reports</i> , 2019, 19, 4711-4718.	1.1	4
77	28 novel mutations identified from 33 Chinese patients with cilia-related kidney disorders. <i>Clinica Chimica Acta</i> , 2020, 501, 207-215.	0.5	4
78	The rare Alu element-mediated chimerism of multiple de novo complex rearrangement sequences in GAN result in giant axonal neuropathy. <i>Clinica Chimica Acta</i> , 2020, 502, 91-98.	0.5	4
79	A novel <i>GJB1</i> mutation associated with X-linked Charcot-Marie-Tooth disease in a large Chinese family pedigree. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1127.	0.6	4
80	IL-24 armored CAR19-T cells show enhanced antitumor activity and persistence. <i>Signal Transduction and Targeted Therapy</i> , 2021, 6, 14.	7.1	4
81	An Episomal CRISPR/Cas12a System for Mediating Efficient Gene Editing. <i>Life</i> , 2021, 11, 1262.	1.1	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. <i>International Journal of Molecular Sciences</i> , 2022, 23, 623.	1.8	4
83	A lesson from a reported pathogenic variant in Peutz-Jeghers syndrome: a case report. <i>Familial Cancer</i> , 2017, 16, 417-422.	0.9	3
84	REDBot: Natural language process methods for clinical copy number variation reporting in prenatal and products of conception diagnosis. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1488.	0.6	3
85	Behavioral and Gene Expression Analysis of Stxbp6-Knockout Mice. <i>Brain Sciences</i> , 2021, 11, 436.	1.1	3
86	Novel variants in OSGEF leading to Galloway-Mowat syndrome by altering its subcellular localization. <i>Clinica Chimica Acta</i> , 2021, 523, 297-303.	0.5	3
87	Identification of four novel mutations in BTK from six Chinese families with X-linked agammaglobulinemia. <i>Clinica Chimica Acta</i> , 2022, 531, 48-55.	0.5	3
88	Cas14a1-Mediated Nucleic Acid Diagnostics for Spinal Muscular Atrophy. <i>Biosensors</i> , 2022, 12, 268.	2.3	3
89	Pre- and postnatal overgrowth in a patient with proximal 4p deletion. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 791-794.	0.7	2
90	Generation of reporter hESCs by targeting <i>EGFP</i> at the CD144 locus to facilitate the endothelial differentiation. <i>Development Growth and Differentiation</i> , 2018, 60, 205-215.	0.6	2

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91	Ectopic expression of factor VIII in MSCs and hepatocytes derived from rDNA targeted hESCs. <i>Clinica Chimica Acta</i> , 2019, 495, 656-663.	0.5	2
92	Identification of pathogenic mutations in 6 Chinese families with multiple exostoses by whole-exome sequencing and multiplex ligation-dependent probe amplification. <i>Medicine (United States)</i> , 2019, 98, e15692.	0.4	2
93	Prenatal Diagnosis in a Fetus With X-Linked Recessive Chondrodysplasia Punctata: Identification and Functional Study of a Novel Missense Mutation in ARSE. <i>Frontiers in Genetics</i> , 2021, 12, 722694.	1.1	2
94	Investigation of hrDNA targeting vector-mediated tumor-specific suicide gene therapy for hepatocellular carcinoma. <i>Science Bulletin</i> , 2006, 51, 2342-2350.	1.7	1
95	A novel FOXL2 mutation in a Chinese family with blepharophimosis, ptosis, epicanthus inversus syndrome. <i>Human Genome Variation</i> , 2015, 2, 15008.	0.4	1
96	Simultaneous Identification of Both MFSD8 and RDH12 Pathogenic Variants in a Chinese Family Affected With Retinitis Pigmentosa. <i>Frontiers in Genetics</i> , 2021, 12, 715100.	1.1	1
97	A patient with apparently reciprocal translocation and cryptic 10p deletion. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1753-1755.	0.7	0
98	Morphometric analysis and developmental comparison of embryos from carriers with balanced chromosomal rearrangements in preimplantation genetic diagnosis cycles. <i>Reproduction, Fertility and Development</i> , 2016, 28, 1953.	0.1	0
99	NovelGZF1pathogenic variants identified in two Chinese patients with Larsen syndrome. <i>Clinical Genetics</i> , 2021, 99, 281-285.	1.0	0
100	Biochemical and genetic characteristics of 40 neonates with carnitine deficiency. <i>Journal of Central South University (Medical Sciences)</i> , 2020, 45, 1164-1171.	0.1	0