# 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 | 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 |
| 2 | 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 |
| 3 | 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 |
| 4 | Comprehensive Analysis of Congenital Adrenal Hyperplasia Using Long-Read Sequencing. Clinical Chemistry, 2022, 68, 927-939. | 3.2 | 23 |
| 5 | Mutation analysis of the TUBB8 gene in primary infertile women with oocyte maturation arrest. Journal of Ovarian Research, 2022, 15, 38. | 3.0 | 14 |
| 6 | 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 |
| 7 | 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 |
| 8 | Cas14a1-Mediated Nucleic Acid Diagnostics for Spinal Muscular Atrophy. Biosensors, 2022, 12, 268. | 4.7 | 3 |
| 9 | NovelGZF1pathogenic variants identified in two Chinese patients with Larsen syndrome. Clinical Genetics, 2021, 99, 281-285. | 2.0 | 0 |
| 10 | IL-24 armored CAR19-T cells show enhanced antitumor activity and persistence. Signal Transduction and Targeted Therapy, 2021, 6, 14. | 17.1 | 4 |
| 11 | 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 |
| 12 | Loss of PIGK function causes severe infantile encephalopathy and extensive neuronal apoptosis. Human Genetics, 2021, 140, 791-803. | 3.8 | 6 |
| 13 | 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 |
| 14 | Behavioral and Gene Expression Analysis of Stxbp6-Knockout Mice. Brain Sciences, 2021, 11, 436. | 2.3 | 3 |
| 15 | 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 |
| 16 | Cas12a and Lateral Flow Strip-Based Test for Rapid and Ultrasensitive Detection of Spinal Muscular Atrophy. Biosensors, 2021, 11, 154. | 4.7 | 9 |
| 17 | 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 |
| 18 | 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 |

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|---|---|---|---|
| 19 | CRISPR/Cas12a-Based Ultrasensitive and Rapid Detection of JAK2 V617F Somatic Mutation in Myeloproliferative Neoplasms. Biosensors, 2021, 11, 247. | 4.7 | 10 |
| 20 | 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 |
| 21 | A More Universal Approach to Comprehensive Analysis of Thalassemia Alleles (CATSA). Journal of Molecular Diagnostics, 2021, 23, 1195-1204. | 2.8 | 55 |
| 22 | 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 |
| 23 | 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 |
| 24 | Novel variants in OSGEP leading to Galloway-Mowat syndrome by altering its subcellular localization. Clinica Chimica Acta, 2021, 523, 297-303. | 1.1 | 3 |
| 25 | An Episomal CRISPR/Cas12a System for Mediating Efficient Gene Editing. Life, 2021, 11, 1262. | 2.4 | 4 |
| 26 | 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 |
| 27 | 28 novel mutations identified from 33 Chinese patients with cilia-related kidney disorders. Clinica Chimica Acta, 2020, 501, 207-215. | 1.1 | 4 |
| 28 | 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 |
| 29 | 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 |
| 30 | 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 |
| 31 | 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 |
| 32 | 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 |
| 33 | 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 |
| 34 | 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 |
| 35 | 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 |
| 36 | Gene Therapy for Hemophilia A: Where We Stand. Current Gene Therapy, 2020, 20, 142-151. | 2.0 | 7 |

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|---|---|---|---|
| 37 | Biochemical and genetic characteristics of 40 neonates with carnitine deficiency. Journal of Central South University (Medical Sciences), 2020, 45, 1164-1171. | 0.1 | 0 |
| 38 | Ectopic expression of factor VIII in MSCs and hepatocytes derived from rDNA targeted hESCs. Clinica Chimica Acta, 2019, 495, 656-663. | 1.1 | 2 |
| 39 | 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 |
| 40 | Molecular genetic study of 59 Chinese Oculocutaneous albinism families. European Journal of Medical Genetics, 2019, 62, 103709. | 1.3 | 5 |
| 41 | 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 |
| 42 | 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 |
| 43 | Clinical utility of noninvasive prenatal screening for expanded chromosome disease syndromes. Genetics in Medicine, 2019, 21, 1998-2006. | 2.4 | 158 |
| 44 | 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 |
| 45 | 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 |
| 46 | 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 |
| 47 | Identification of Five Novel Mutations Causing Rare Lysosomal Storage Diseases. Medical Science Monitor, 2019, 25, 7634-7644. | 1.1 | 5 |
| 48 | Gene Therapy for Hemophilia and Duchenne Muscular Dystrophy in China. Human Gene Therapy, 2018, 29, 146-150. | 2.7 | 5 |
| 49 | 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 |
| 50 | 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 |
| 51 | 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 |
| 52 | 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 |
| 53 | Constructing a database for the relations between CNV and human genetic diseases via systematic text mining. BMC Bioinformatics, 2018, 19, 528. | 2.6 | 16 |
| 54 | 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 |

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|---|---|---|---|
| 55 | 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 |
| 56 | 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 |
| 57 | 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 |
| 58 | 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 |
| 59 | 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 |
| 60 | Novel GATAD2B loss-of-function mutations cause intellectual disability in two unrelated cases. Journal of Human Genetics, 2017, 62, 513-516. | 2.3 | 12 |
| 61 | A lesson from a reported pathogenic variant in Peutz-Jeghers syndrome: a case report. Familial Cancer, 2017, 16, 417-422. | 1.9 | 3 |
| 62 | 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 |
| 63 | 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 |
| 64 | 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 |
| 65 | 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 |
| 66 | 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 |
| 67 | 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 |
| 68 | 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 |
| 69 | In situ genetic correction of F8 intron 22 inversion in hemophilia A patient-specific iPSCs. Scientific Reports, 2016, 6, 18865. | 3.3 | 43 |
| 70 | 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 |
| 71 | 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 |
| 72 | Three novel mutations of STK11 gene in Chinese patients with Peutzâ€“Jeghers syndrome. BMC Medical Genetics, 2016, 17, 77. | 2.1 | 8 |

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|---|---|---|---|
| 73 | A novel de novo POGZ mutation in a patient with intellectual disability. Journal of Human Genetics, 2016, 61, 357-359. | 2.3 | 31 |
| 74 | Novel and reported APC germline mutations in Chinese patients with familial adenomatous polyposis. Gene, 2016, 577, 187-192. | 2.2 | 11 |
| 75 | Clinical and molecular investigation in Chinese patients with glutaric aciduria type I. Clinica Chimica Acta, 2016, 453, 75-79. | 1.1 | 11 |
| 76 | A novel FOXL2 mutation in a Chinese family with blepharophimosis, ptosis, epicanthus inversus syndrome. Human Genome Variation, 2015, 2, 15008. | 0.7 | 1 |
| 77 | 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 |
| 78 | Eight-Shaped Hatching Increases the Risk of Inner Cell Mass Splitting in Extended Mouse Embryo Culture. PLoS ONE, 2015, 10, e0145172. | 2.5 | 19 |
| 79 | 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 |
| 80 | Rare intracranial cholesterol deposition and a homozygous mutation of LDLR in a familial hypercholesterolemia patient. Gene, 2015, 569, 313-317. | 2.2 | 5 |
| 81 | 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 |
| 82 | 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 |
| 83 | Partial trisomy 2q33.3-q37.3 in a patient with an inverted duplicated neocentric marker chromosome. Molecular Cytogenetics, 2015, 8, 10. | 0.9 | 4 |
| 84 | DMD mutation spectrum analysis in 613 Chinese patients with dystrophinopathy. Journal of Human Genetics, 2015, 60, 435-442. | 2.3 | 47 |
| 85 | 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 |
| 86 | Two novel NIPBL gene mutations in Chinese patients with Cornelia de Lange syndrome. Gene, 2015, 555, 476-480. | 2.2 | 5 |
| 87 | 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 |
| 88 | 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 |
| 89 | 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 |
| 90 | 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 |

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|---|---|---|---|
| 91 | Copy Number Variation Sequencing for Comprehensive Diagnosis of Chromosome Disease Syndromes. Journal of Molecular Diagnostics, 2014, 16, 519-526. | 2.8 | 106 |
| 92 | 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 |
| 93 | A patient with apparently reciprocal translocation and cryptic 10p deletion. American Journal of Medical Genetics, Part A, 2011, 155, 1753-1755. | 1.2 | 0 |
| 94 | 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 |
| 95 | A ZRS duplication causes syndactyly type IV with tibial hypoplasia. American Journal of Medical Genetics, Part A, 2009, 149A, 816-818. | 1.2 | 35 |
| 96 | 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 |
| 97 | 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 |
| 98 | Investigation of hrDNA targeting vector-mediated tumor-specific suicide gene therapy for hepatocellular carcinoma. Science Bulletin, 2006, 51, 2342-2350. | 1.7 | 1 |
| 99 | Expression of reconstructive hFVIII in the hrDNA by using hrDNA targeting vector. Science Bulletin, 2005, 50, 2187-2192. | 1.7 | 4 |
| 100 | Silica nanoparticle is a possible safe carrier for gene therapy. Science Bulletin, 2005, 50, 2323-2327. | 1.7 | 10 |