

Kun Xia

List of Publications by Year in descending order

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Version: 2024-02-01

223
papers

6,942
citations

76326
40
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98798
67
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234
all docs

234
docs citations

234
times ranked

10986
citing authors

| # | ARTICLE | IF | CITATIONS |
|----|--|------|-----------|
| 1 | Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. <i>Nature Genetics</i> , 2017, 49, 515-526. | 21.4 | 443 |
| 2 | De novo genic mutations among a Chinese autism spectrum disorder cohort. <i>Nature Communications</i> , 2016, 7, 13316. | 12.8 | 293 |
| 3 | Association of PINK1 and DJ-1 confers digenic inheritance of early-onset Parkinson's disease. <i>Human Molecular Genetics</i> , 2006, 15, 1816-1825. | 2.9 | 218 |
| 4 | Expansion of Human-Specific GGC Repeat in Neuronal Intranuclear Inclusion Disease-Related Disorders. <i>American Journal of Human Genetics</i> , 2019, 105, 166-176. | 6.2 | 212 |
| 5 | Prevalence of Autism Spectrum Disorder in China: A Nationwide Multi-center Population-based Study Among Children Aged 6 to 12 Years. <i>Neuroscience Bulletin</i> , 2020, 36, 961-971. | 2.9 | 179 |
| 6 | Genes with de novo mutations are shared by four neuropsychiatric disorders discovered from NPdenovo database. <i>Molecular Psychiatry</i> , 2016, 21, 290-297. | 7.9 | 167 |
| 7 | Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. <i>Nature Neuroscience</i> , 2017, 20, 1043-1051. | 14.8 | 152 |
| 8 | AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. <i>Nature Communications</i> , 2019, 10, 3094. | 12.8 | 150 |
| 9 | Expansion of GGC repeat in the human-specific NOTCH2NLC gene is associated with essential tremor. <i>Brain</i> , 2020, 143, 222-233. | 7.6 | 139 |
| 10 | Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. <i>American Journal of Human Genetics</i> , 2016, 98, 541-552. | 6.2 | 132 |
| 11 | Ångstrom-scale silver particle“embedded carbomer gel promotes wound healing by inhibiting bacterial colonization and inflammation. <i>Science Advances</i> , 2020, 6, . | 10.3 | 119 |
| 12 | Inherited and multiple de novo mutations in autism/developmental delay risk genes suggest a multifactorial model. <i>Molecular Autism</i> , 2018, 9, 64. | 4.9 | 114 |
| 13 | Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. <i>Nature Communications</i> , 2020, 11, 4932. | 12.8 | 105 |
| 14 | The role of genetics in Parkinson's disease: a large cohort study in Chinese mainland population. <i>Brain</i> , 2020, 143, 2220-2234. | 7.6 | 97 |
| 15 | Targeted sequencing and functional analysis reveal brain-size-related genes and their networks in autism spectrum disorders. <i>Molecular Psychiatry</i> , 2017, 22, 1282-1290. | 7.9 | 95 |
| 16 | Excessive UBE3A dosage impairs retinoic acid signaling and synaptic plasticity in autism spectrum disorders. <i>Cell Research</i> , 2018, 28, 48-68. | 12.0 | 95 |
| 17 | Genetic evidence of gender difference in autism spectrum disorder supports the female-protective effect. <i>Translational Psychiatry</i> , 2020, 10, 4. | 4.8 | 84 |
| 18 | <i>SLC39A5</i> mutations interfering with the BMP/TGF- β 2 pathway in non-syndromic high myopia. <i>Journal of Medical Genetics</i> , 2014, 51, 518-525. | 3.2 | 83 |

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|----|--|------|-----------|
| 19 | Long-read sequencing identified intronic repeat expansions in <i>SAMD12</i> from Chinese pedigrees affected with familial cortical myoclonic tremor with epilepsy. <i>Journal of Medical Genetics</i> , 2019, 56, 265-270. | 3.2 | 82 |
| 20 | Coding mutations in <i>NUS1</i> contribute to Parkinson's disease. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, 11567-11572. | 7.1 | 78 |
| 21 | miR-222 attenuates cisplatin-induced cell death by targeting the <i>PPP2R2A/Akt/mTOR</i> Axis in bladder cancer cells. <i>Journal of Cellular and Molecular Medicine</i> , 2016, 20, 559-567. | 3.6 | 74 |
| 22 | A <i>SIGMAR1</i> splice-site mutation causes distal hereditary motor neuropathy. <i>Neurology</i> , 2015, 84, 2430-2437. | 1.1 | 72 |
| 23 | Inhibition of miR-331-3p and miR-9-5p ameliorates Alzheimer's disease by enhancing autophagy. <i>Theranostics</i> , 2021, 11, 2395-2409. | 10.0 | 72 |
| 24 | Extracellular vesicles from human urine-derived stem cells prevent osteoporosis by transferring <i>CTHRC1</i> and <i>OPG</i> . <i>Bone Research</i> , 2019, 7, 18. | 11.4 | 66 |
| 25 | <i>PAK2</i> Haploinsufficiency Results in Synaptic Cytoskeleton Impairment and Autism-Related Behavior. <i>Cell Reports</i> , 2018, 24, 2029-2041. | 6.4 | 64 |
| 26 | OncoVar: an integrated database and analysis platform for oncogenic driver variants in cancers. <i>Nucleic Acids Research</i> , 2021, 49, D1289-D1301. | 14.5 | 64 |
| 27 | Mutations of <i>P4HA2</i> encoding prolyl 4-hydroxylase 2 are associated with nonsyndromic high myopia. <i>Genetics in Medicine</i> , 2015, 17, 300-306. | 2.4 | 63 |
| 28 | Autophagy receptor <i>OPTN</i> (optineurin) regulates mesenchymal stem cell fate and bone-fat balance during aging by clearing <i>FABP3</i> . <i>Autophagy</i> , 2021, 17, 2766-2782. | 9.1 | 63 |
| 29 | Mutation analysis of <i>Parkin</i> , <i>PINK1</i> , <i>DJ-1</i> and <i>ATP13A2</i> genes in Chinese patients with autosomal recessive early-onset Parkinsonism. <i>Movement Disorders</i> , 2008, 23, 2074-2079. | 3.9 | 61 |
| 30 | Novel West syndrome candidate genes in a Chinese cohort. <i>CNS Neuroscience and Therapeutics</i> , 2018, 24, 1196-1206. | 3.9 | 60 |
| 31 | Analysis of lncRNAs expression in UVB-induced stress responses of melanocytes. <i>Journal of Dermatological Science</i> , 2016, 81, 53-60. | 1.9 | 57 |
| 32 | Elevated mitochondrial DNA copy number in peripheral blood cells is associated with childhood autism. <i>BMC Psychiatry</i> , 2015, 15, 50. | 2.6 | 56 |
| 33 | Identification of <i>C9orf72</i> repeat expansions in patients with amyotrophic lateral sclerosis and frontotemporal dementia in mainland China. <i>Neurobiology of Aging</i> , 2014, 35, 936.e19-936.e22. | 3.1 | 53 |
| 34 | Glucocerebrosidase Gene L444P mutation is a risk factor for Parkinson's disease in Chinese population. <i>Movement Disorders</i> , 2010, 25, 1005-1011. | 3.9 | 50 |
| 35 | Investigation of Gene Regulatory Networks Associated with Autism Spectrum Disorder Based on MiRNA Expression in China. <i>PLoS ONE</i> , 2015, 10, e0129052. | 2.5 | 50 |
| 36 | Genome-wide copy number variation analysis in a Chinese autism spectrum disorder cohort. <i>Scientific Reports</i> , 2017, 7, 44155. | 3.3 | 50 |

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|----|---|------|-----------|
| 37 | Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. <i>Genome Medicine</i> , 2021, 13, 63. | 8.2 | 50 |
| 38 | Extracellular Vesicles from <i>Akkermansia muciniphila</i> Elicit Antitumor Immunity Against Prostate Cancer via Modulation of CD8+ T Cells and Macrophages. <i>International Journal of Nanomedicine</i> , 2021, Volume 16, 2949-2963. | 6.7 | 48 |
| 39 | Comparing the benefits of chemoradiotherapy and chemotherapy for resectable stage III A/N2 non-small cell lung cancer: a meta-analysis. <i>World Journal of Surgical Oncology</i> , 2018, 16, 8. | 1.9 | 47 |
| 40 | Fasting before or after wound injury accelerates wound healing through the activation of pro-angiogenic SMOC1 and SCG2. <i>Theranostics</i> , 2020, 10, 3779-3792. | 10.0 | 44 |
| 41 | Analysis of SCA2 and SCA3/MJD repeats in Parkinson's disease in mainland China: Genetic, clinical, and positron emission tomography findings. <i>Movement Disorders</i> , 2009, 24, 2007-2011. | 3.9 | 43 |
| 42 | <i>AKAP2</i> identified as a novel gene mutated in a Chinese family with adolescent idiopathic scoliosis. <i>Journal of Medical Genetics</i> , 2016, 53, 488-493. | 3.2 | 43 |
| 43 | Familial paroxysmal kinesigenic dyskinesia is associated with mutations in the KCNA1 gene. <i>Human Molecular Genetics</i> , 2018, 27, 625-637. | 2.9 | 43 |
| 44 | Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. <i>Nature Communications</i> , 2019, 10, 4679. | 12.8 | 43 |
| 45 | Extracellular vesicles from human umbilical cord blood ameliorate bone loss in senile osteoporotic mice. <i>Metabolism: Clinical and Experimental</i> , 2019, 95, 93-101. | 3.4 | 43 |
| 46 | Suppression of Akt-mTOR pathway rescued the social behavior in <i>Cntnap2</i> -deficient mice. <i>Scientific Reports</i> , 2019, 9, 3041. | 3.3 | 43 |
| 47 | Alterations of the Gut Microbiota in Multiple System Atrophy Patients. <i>Frontiers in Neuroscience</i> , 2019, 13, 1102. | 2.8 | 42 |
| 48 | Gene4Denovo: an integrated database and analytic platform for de novo mutations in humans. <i>Nucleic Acids Research</i> , 2020, 48, D913-D926. | 14.5 | 41 |
| 49 | Biallelic Intronic AAGGG Expansion of <i>RFC1</i> is Related to Multiple System Atrophy. <i>Annals of Neurology</i> , 2020, 88, 1132-1143. | 5.3 | 41 |
| 50 | Increased Expression of Reticulon 3 in Neurons Leads to Reduced Axonal Transport of β Site Amyloid Precursor Protein-cleaving Enzyme 1. <i>Journal of Biological Chemistry</i> , 2013, 288, 30236-30245. | 3.4 | 40 |
| 51 | 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. | 2.3 | 39 |
| 52 | Leukocyte Mitochondrial DNA Copy Number in Blood Is Not Associated with Major Depressive Disorder in Young Adults. <i>PLoS ONE</i> , 2014, 9, e96869. | 2.5 | 39 |
| 53 | Association of genetic variants of GRIN2B with autism. <i>Scientific Reports</i> , 2015, 5, 8296. | 3.3 | 39 |
| 54 | Fluorometric aptamer-based determination of ochratoxin A based on the use of graphene oxide and RNase H-aided amplification. <i>Mikrochimica Acta</i> , 2018, 185, 347. | 5.0 | 39 |

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|----|--|------|-----------|
| 55 | Methylation analysis of plasma cell-free DNA for breast cancer early detection using bisulfite next-generation sequencing. <i>Tumor Biology</i> , 2016, 37, 13111-13119. | 1.8 | 38 |
| 56 | miR-25 alleviates polyQ-mediated cytotoxicity by silencing <i>ATXN3</i> . <i>FEBS Letters</i> , 2014, 588, 4791-4798. | 2.8 | 37 |
| 57 | (CAG) ⁿ loci as genetic modifiers of age-at-onset in patients with Machado-Joseph disease from mainland China. <i>Brain</i> , 2016, 139, e41-e41. | 7.6 | 37 |
| 58 | Fructose-coated Angstrom silver inhibits osteosarcoma growth and metastasis via promoting ROS-dependent apoptosis through the alteration of glucose metabolism by inhibiting PDK. <i>Theranostics</i> , 2020, 10, 7710-7729. | 10.0 | 37 |
| 59 | A syndactyly type IV locus maps to 7q36. <i>Journal of Human Genetics</i> , 2007, 52, 561-564. | 2.3 | 35 |
| 60 | mirTrios: an integrated pipeline for detection of de novo and rare inherited mutations from trios-based next-generation sequencing. <i>Journal of Medical Genetics</i> , 2015, 52, 275-281. | 3.2 | 35 |
| 61 | Altered plasma levels of chemokines in autism and their association with social behaviors. <i>Psychiatry Research</i> , 2016, 244, 300-305. | 3.3 | 35 |
| 62 | Disruptive variants of <i>CSDE1</i> associate with autism and interfere with neuronal development and synaptic transmission. <i>Science Advances</i> , 2019, 5, eaax2166. | 10.3 | 35 |
| 63 | Shorter telomere length in peripheral blood leukocytes is associated with childhood autism. <i>Scientific Reports</i> , 2014, 4, 7073. | 3.3 | 34 |
| 64 | Neuronal Induction of Bone-Fat Imbalance through Osteocyte Neuropeptide Y. <i>Advanced Science</i> , 2021, 8, e2100808. | 11.2 | 34 |
| 65 | Updated frequency analysis of spinocerebellar ataxia in China. <i>Brain</i> , 2018, 141, e22-e22. | 7.6 | 33 |
| 66 | Genomic landscapes of Chinese sporadic autism spectrum disorders revealed by whole-genome sequencing. <i>Journal of Genetics and Genomics</i> , 2018, 45, 527-538. | 3.9 | 33 |
| 67 | The repertoire of tumor-infiltrating lymphocytes within the microenvironment of oral squamous cell carcinoma reveals immune dysfunction. <i>Cancer Immunology, Immunotherapy</i> , 2020, 69, 465-476. | 4.2 | 32 |
| 68 | Screening for distress in patients with primary brain tumor using distress thermometer: a systematic review and meta-analysis. <i>BMC Cancer</i> , 2018, 18, 124. | 2.6 | 31 |
| 69 | Ångstrom-Scale Silver Particles as a Promising Agent for Low-Toxicity Broad-Spectrum Potent Anticancer Therapy. <i>Advanced Functional Materials</i> , 2019, 29, 1808556. | 14.9 | 29 |
| 70 | Phenotype-to-genotype approach reveals head-circumference-associated genes in an autism spectrum disorder cohort. <i>Clinical Genetics</i> , 2020, 97, 338-346. | 2.0 | 29 |
| 71 | Increased Reticulon 3 (RTN3) Leads to Obesity and Hypertriglyceridemia by Interacting With Heat Shock Protein Family A (Hsp70) Member 5 (HSPA5). <i>Circulation</i> , 2018, 138, 1828-1838. | 1.6 | 26 |
| 72 | A Statistical Framework for Mapping Risk Genes from De Novo Mutations in Whole-Genome-Sequencing Studies. <i>American Journal of Human Genetics</i> , 2018, 102, 1031-1047. | 6.2 | 26 |

| # | ARTICLE | IF | CITATIONS |
|----|--|-----|-----------|
| 73 | The Prognostic Value of Altered eIF3a and Its Association with p27 in Non-Small Cell Lung Cancers. PLoS ONE, 2014, 9, e96008. | 2.5 | 26 |
| 74 | The APOE ϵ 2 allele may decrease the age at onset in patients with spinocerebellar ataxia type 3 or Machado-Joseph disease from the Chinese Han population. Neurobiology of Aging, 2014, 35, 2179.e15-2179.e18. | 3.1 | 25 |
| 75 | Exome sequencing identifies POU4F3 as the causative gene for a large Chinese family with non-syndromic hearing loss. Journal of Human Genetics, 2017, 62, 317-320. | 2.3 | 25 |
| 76 | Genotype and phenotype correlations for <i>SHANK3</i> de novo mutations in neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2018, 176, 2668-2676. | 1.2 | 25 |
| 77 | A novel PRPF31 splice-site mutation in a Chinese family with autosomal dominant retinitis pigmentosa. Molecular Vision, 2004, 10, 361-5. | 1.1 | 25 |
| 78 | De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. Genetics in Medicine, 2020, 22, 538-546. | 2.4 | 24 |
| 79 | A Novel Mutation of <i>FOXC1</i> (R127L) in an Axenfeld-Rieger Syndrome Family with Glaucoma and Multiple Congenital Heart Diseases. Ophthalmic Genetics, 2016, 37, 1-5. | 1.2 | 23 |
| 80 | Identification of a potential exosomal biomarker in spinocerebellar ataxia Type 3/Machado-Joseph disease. Epigenomics, 2019, 11, 1037-1056. | 2.1 | 23 |
| 81 | An adaptive immune response driven by mature, antigen-experienced <i>T</i> and <i>B</i> cells within the microenvironment of oral squamous cell carcinoma. International Journal of Cancer, 2016, 138, 2952-2962. | 5.1 | 22 |
| 82 | The genetic spectrum of familial hypercholesterolemia in the central south region of China. Atherosclerosis, 2017, 258, 84-88. | 0.8 | 22 |
| 83 | Identification of rare RTN3 variants in Alzheimer's disease in Han Chinese. Human Genetics, 2018, 137, 141-150. | 3.8 | 22 |
| 84 | A comparative study of the genetic components of three subcategories of autism spectrum disorder. Molecular Psychiatry, 2019, 24, 1720-1731. | 7.9 | 22 |
| 85 | Variation of global DNA methylation levels with age and in autistic children. Human Genomics, 2016, 10, 31. | 2.9 | 21 |
| 86 | Rare GCH1 heterozygous variants contributing to Parkinson's disease. Brain, 2017, 140, e41-e41. | 7.6 | 21 |
| 87 | Identification of TDP-43 as an oncogene in melanoma and its function during melanoma pathogenesis. Cancer Biology and Therapy, 2017, 18, 8-15. | 3.4 | 21 |
| 88 | Gene-Related Cerebellar Neurodegeneration in SCA3/MJD: A Case-Controlled Imaging-Genetic Study. Frontiers in Neurology, 2019, 10, 1025. | 2.4 | 21 |
| 89 | H-type blood vessels participate in alveolar bone remodeling during murine tooth extraction healing. Oral Diseases, 2020, 26, 998-1009. | 3.0 | 21 |
| 90 | miR-199a-5p regulates the expression of metastasis-associated genes in B16F10 melanoma cells. International Journal of Clinical and Experimental Pathology, 2014, 7, 7182-90. | 0.5 | 21 |

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|-----|---|------|-----------|
| 91 | Identification of a de novo DYNC1H1 mutation via WES according to published guidelines. Scientific Reports, 2016, 6, 20423. | 3.3 | 20 |
| 92 | Vitamin D-related genes are subjected to significant <i>de novo</i> mutation burdens in autism spectrum disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2017, 174, 568-577. | 1.7 | 20 |
| 93 | Association Between Vitamins and Amyotrophic Lateral Sclerosis: A Center-Based Survey in Mainland China. Frontiers in Neurology, 2020, 11, 488. | 2.4 | 20 |
| 94 | Two Novel SNPs in ATXN3 3' UTR May Decrease Age at Onset of SCA3/MJD in Chinese Patients. PLoS ONE, 2015, 10, e0117488. | 2.5 | 19 |
| 95 | Insertion of a knockout-first cassette in Ampd1 gene leads to neonatal death by disruption of neighboring genes expression. Scientific Reports, 2016, 6, 35970. | 3.3 | 19 |
| 96 | Atoh7 promotes retinal Müller cell differentiation into retinal ganglion cells. Cytotechnology, 2016, 68, 267-277. | 1.6 | 19 |
| 97 | A <i>de novo</i> mutation of <i>SMYD1</i> (p.F272L) is responsible for hypertrophic cardiomyopathy in a Chinese patient. Clinical Chemistry and Laboratory Medicine, 2019, 57, 532-539. | 2.3 | 19 |
| 98 | Alteration of methylation status in the ATXN3 gene promoter region is linked to the SCA3/MJD. Neurobiology of Aging, 2017, 53, 192.e5-192.e10. | 3.1 | 18 |
| 99 | NCKAP1 Disruptive Variants Lead to a Neurodevelopmental Disorder with Core Features of Autism. American Journal of Human Genetics, 2020, 107, 963-976. | 6.2 | 18 |
| 100 | Trafficking abnormality and ER stress underlie functional deficiency of hearing impairment-associated connexin-31 mutants. Protein and Cell, 2010, 1, 935-943. | 11.0 | 17 |
| 101 | Roles of KChIP1 in the regulation of GABA-mediated transmission and behavioral anxiety. Molecular Brain, 2010, 3, 23. | 2.6 | 17 |
| 102 | MFN2-related genetic and clinical features in a cohort of Chinese CMT2 patients. Journal of the Peripheral Nervous System, 2016, 21, 38-44. | 3.1 | 17 |
| 103 | A turn-on fluorescence assay of alkaline phosphatase activity using a DNA-silver nanocluster probe. New Journal of Chemistry, 2018, 42, 4331-4336. | 2.8 | 17 |
| 104 | Rare inherited missense variants of POGZ associate with autism risk and disrupt neuronal development. Journal of Genetics and Genomics, 2019, 46, 247-257. | 3.9 | 17 |
| 105 | Is the High Frequency of Machado-Joseph Disease in China Due to New Mutational Origins?. Frontiers in Genetics, 2018, 9, 740. | 2.3 | 17 |
| 106 | A novel fusion suicide gene yeast CDglyTK plays a role in radio-gene therapy of nasopharyngeal carcinoma. Cancer Gene Therapy, 2004, 11, 790-796. | 4.6 | 16 |
| 107 | Label-free fluorescence turn-on detection of uracil DNA glycosylase activity based on G-quadruplex formation. Talanta, 2016, 160, 449-453. | 5.5 | 16 |
| 108 | <i>ATXN2</i> polymorphism modulates age at onset in Machado-Joseph disease. Brain, 2016, 139, aww176. | 7.6 | 16 |

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|-----|---|-----|-----------|
| 109 | Association of <i>TNFRSF10A</i> rs1799964 and <i>IL12A</i> rs16944 polymorphisms with multiple system atrophy in Chinese Han population. <i>International Journal of Neuroscience</i> , 2018, 128, 761-764. | 1.6 | 16 |
| 110 | Genome-wide association analysis of autism identified multiple loci that have been reported as strong signals for neuropsychiatric disorders. <i>Autism Research</i> , 2020, 13, 382-396. | 3.8 | 16 |
| 111 | Hmgb1 inhibits Klotho expression and malignant phenotype in melanoma cells by activating NF- κ B. <i>Oncotarget</i> , 2016, 7, 80765-80782. | 1.8 | 16 |
| 112 | A Novel Alzheimer-Associated SNP in Tmp21 Increases Amyloidogenesis. <i>Molecular Neurobiology</i> , 2018, 55, 1862-1870. | 4.0 | 15 |
| 113 | Sleep Problems of Children with Autism May Independently Affect Parental Quality of Life. <i>Child Psychiatry and Human Development</i> , 2021, 52, 488-499. | 1.9 | 15 |
| 114 | Parecoxib inhibits glioblastoma cell proliferation, migration and invasion by up-regulating miRNA-29c. <i>Biology Open</i> , 2017, 6, 311-316. | 1.2 | 14 |
| 115 | IGHMBP2-related clinical and genetic features in a cohort of Chinese Charcot-Marie-Tooth disease type 2 patients. <i>Neuromuscular Disorders</i> , 2017, 27, 193-199. | 0.6 | 14 |
| 116 | Genotype-phenotype correlation and frequency of distribution in a cohort of Chinese Charcot-Marie-Tooth patients associated with GDAP1 mutations. <i>Journal of Neurology</i> , 2018, 265, 637-646. | 3.6 | 14 |
| 117 | Pathogenic variants in <i>TNRC6B</i> cause a genetic disorder characterised by developmental delay/intellectual disability and a spectrum of neurobehavioural phenotypes including autism and ADHD. <i>Journal of Medical Genetics</i> , 2020, 57, 717-724. | 3.2 | 14 |
| 118 | Autism spectrum disorder and severe social impairment associated with elevated plasma interleukin-8. <i>Pediatric Research</i> , 2021, 89, 591-597. | 2.3 | 14 |
| 119 | Biallelic loss-of-function variants in NEMF cause central nervous system impairment and axonal polyneuropathy. <i>Human Genetics</i> , 2021, 140, 579-592. | 3.8 | 14 |
| 120 | Ag nanoparticles enhance immune checkpoint blockade efficacy by promoting of immune surveillance in melanoma. <i>Journal of Colloid and Interface Science</i> , 2022, 616, 189-200. | 9.4 | 14 |
| 121 | Prediction of orthostatic hypotension in multiple system atrophy and Parkinson disease. <i>Scientific Reports</i> , 2016, 6, 21649. | 3.3 | 13 |
| 122 | Birt-Hogg-Dubé syndrome in two Chinese families with mutations in the FLCN gene. <i>BMC Medical Genetics</i> , 2018, 19, 14. | 2.1 | 13 |
| 123 | POGZ de novo missense variants in neuropsychiatric disorders. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e900. | 1.2 | 13 |
| 124 | G3BP1 may serve as a potential biomarker of proliferation, apoptosis, and prognosis in oral squamous cell carcinoma. <i>Journal of Oral Pathology and Medicine</i> , 2021, 50, 995-1004. | 2.7 | 13 |
| 125 | iTRAQ-Based Quantitative Proteomic Analysis of Nasopharyngeal Carcinoma. <i>Journal of Cellular Biochemistry</i> , 2015, 116, 1431-1441. | 2.6 | 12 |
| 126 | GLA variation p.E66Q identified as the genetic etiology of Fabry disease using exome sequencing. <i>Gene</i> , 2016, 575, 363-367. | 2.2 | 12 |

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|-----|--|-----|-----------|
| 127 | Novel mutations in ADSL for Adenylosuccinate Lyase Deficiency identified by the combination of Trio-WES and constantly updated guidelines. Scientific Reports, 2017, 7, 1625. | 3.3 | 12 |
| 128 | Homemade-device-induced negative pressure promotes wound healing more efficiently than VSD-induced positive pressure by regulating inflammation, proliferation and remodeling. International Journal of Molecular Medicine, 2017, 39, 879-888. | 4.0 | 12 |
| 129 | A novel NHS mutation causes Nance-Horan Syndrome in a Chinese family. BMC Medical Genetics, 2017, 18, 2. | 2.1 | 12 |
| 130 | Metformin reduces the increased risk of oral squamous cell carcinoma recurrence in patients with type 2 diabetes mellitus: A cohort study with propensity score analyses. Surgical Oncology, 2020, 35, 453-459. | 1.6 | 12 |
| 131 | Assessment of causal effects of physical activity on neurodegenerative diseases: A Mendelian randomization study. Journal of Sport and Health Science, 2021, 10, 454-461. | 6.5 | 12 |
| 132 | Analysis of the GGGGCC Repeat Expansions of the C9orf72 Gene in SCA3/MJD Patients from China. PLoS ONE, 2015, 10, e0130336. | 2.5 | 11 |
| 133 | Carnosic acid attenuates acute ethanol-induced liver injury via a SIRT1/p66Shc-mediated mitochondrial pathway. Canadian Journal of Physiology and Pharmacology, 2016, 94, 416-425. | 1.4 | 11 |
| 134 | Prediction of the Age at Onset of Spinocerebellar Ataxia Type 3 with Machine Learning. Movement Disorders, 2021, 36, 216-224. | 3.9 | 11 |
| 135 | Targeted sequencing and integrative analysis of 3,195 Chinese patients with neurodevelopmental disorders prioritized 26 novel candidate genes. Journal of Genetics and Genomics, 2021, 48, 312-323. | 3.9 | 11 |
| 136 | A novel transgenic mouse model of Chinese Charcot-Marie-Tooth disease type 2L. Neural Regeneration Research, 2014, 9, 413. | 3.0 | 11 |
| 137 | A novel defined hypoxia-related gene signature to predict the prognosis of oral squamous cell carcinoma. Annals of Translational Medicine, 2021, 9, 1565-1565. | 1.7 | 11 |
| 138 | Silica nanoparticle is a possible safe carrier for gene therapy. Science Bulletin, 2005, 50, 2323-2327. | 1.7 | 10 |
| 139 | Ubiquitin-related network underlain by (CAC) _n loci modulate age at onset in Machado-Joseph disease. Brain, 2017, 140, e25-e25. | 7.6 | 10 |
| 140 | CD147 promotes proliferation and migration of oral cancer cells by inhibiting junctions between E-cadherin and β -catenin. Journal of Oral Pathology and Medicine, 2020, 49, 1019-1029. | 2.7 | 10 |
| 141 | Common genetic variants shared among five major psychiatric disorders: a large-scale genome-wide combined analysis. Global Clinical and Translational Research, 2019, , 21-30. | 0.3 | 10 |
| 142 | New ZNF644 mutations identified in patients with high myopia. Molecular Vision, 2014, 20, 939-46. | 1.1 | 10 |
| 143 | Genetic Diagnosis of Two Dopa-Responsive Dystonia Families by Exome Sequencing. PLoS ONE, 2014, 9, e106388. | 2.5 | 9 |
| 144 | Association and gene-gene interactions study of reelin signaling pathway related genes with autism in the Chinese population. Autism Research, 2016, 9, 436-442. | 3.8 | 9 |

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|-----|---|-----|-----------|
| 145 | Pathogenic missense mutation pattern of forkhead box genes in neurodevelopmental disorders. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e00789. | 1.2 | 9 |
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