Kun Xia

List of Publications by Year
in descending order

223	6,942	76196	98622
papers	citations	40 h-index	67 g-index

234	234	234	10986
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Performance Comparison of Computational Methods for the Prediction of the Function and Pathogenicity of Non-Coding Variants. Genomics, Proteomics and Bioinformatics, 2023, 21, 649-661.	3.0	7
2	GLRA2gene mutations cause high myopia in humans and mice. Journal of Medical Genetics, 2023, 60, 193-203.	1.5	5
3	Study on the expression and function of chordinâ€like 1 in oral squamous cell carcinoma. Oral Diseases, 2023, 29, 2034-2051.	1.5	2
4	Cross-Disorder Analysis of De Novo Mutations in Neuropsychiatric Disorders. Journal of Autism and Developmental Disorders, 2022, 52, 1299-1313.	1.7	3
5	Identification of the Largest SCA36 Pedigree in Asia: with Multimodel Neuroimaging Evaluation for the First Time. Cerebellum, 2022, 21, 358-367.	1.4	3
6	Differentiated embryo chondrocyte 1, induced by hypoxia-inducible factor 1Î±, promotes cell migration in oral squamous cell carcinoma cell lines. Oral Surgery, Oral Medicine, Oral Pathology and Oral Radiology, 2022, 133, 199-206.	0.2	3
7	Blood Neurofilament Light Chain in Genetic Ataxia: A Metaâ€Analysis. Movement Disorders, 2022, 37, 171-181.	2.2	8
8	Type 2 diabetes mellitus promotes the proliferation, metastasis, and suppresses the apoptosis in oral squamous cell carcinoma. Journal of Oral Pathology and Medicine, 2022, 51, 483-492.	1.4	7
9	Loss-of-function of KMT5B leads to neurodevelopmental disorder and impairs neuronal development and neurogenesis. Journal of Genetics and Genomics, 2022, 49, 881-890.	1.7	7
10	Odor identification impairment in autism spectrum disorder might be associated with mitochondrial dysfunction. Asian Journal of Psychiatry, 2022, 72, 103072.	0.9	5
11	Ag nanoparticles enhance immune checkpoint blockade efficacy by promoting of immune surveillance in melanoma. Journal of Colloid and Interface Science, 2022, 616, 189-200.	5.0	14
12	Genetic landscape of human mitochondrial genome using whole-genome sequencing. Human Molecular Genetics, 2022, 31, 1747-1761.	1.4	4
13	The progression rate of spinocerebellar ataxia type 3 varies with disease stage. Journal of Translational Medicine, 2022, 20, 226.	1.8	5
14	Autophagy receptor OPTN (optineurin) regulates mesenchymal stem cell fate and bone-fat balance during aging by clearing FABP3. Autophagy, 2021, 17, 2766-2782.	4.3	63
15	Autism spectrum disorder and severe social impairment associated with elevated plasma interleukin-8. Pediatric Research, 2021, 89, 591-597.	1.1	14
16	Sleep Problems of Children with Autism May Independently Affect ParentalÂQuality of Life. Child Psychiatry and Human Development, 2021, 52, 488-499.	1.1	15
17	Prediction of the Age at Onset of Spinocerebellar Ataxia Type 3 with Machine Learning. Movement Disorders, 2021, 36, 216-224.	2.2	11
18	Biallelic loss-of-function variants in NEMF cause central nervous system impairment and axonal polyneuropathy. Human Genetics, 2021, 140, 579-592.	1.8	14

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19	OncoVar: an integrated database and analysis platform for oncogenic driver variants in cancers. Nucleic Acids Research, 2021, 49, D1289-D1301.	6.5	64
20	Inhibition of miR-331-3p and miR-9-5p ameliorates Alzheimer's disease by enhancing autophagy. Theranostics, 2021, 11, 2395-2409.	4.6	72
21	GPCards: An integrated database of genotypeâ€“phenotype correlations in human genetic diseases. Computational and Structural Biotechnology Journal, 2021, 19, 1603-1611.	1.9	5
22	De novo mutations in folate-related genes associated with common developmental disorders. Computational and Structural Biotechnology Journal, 2021, 19, 1414-1422.	1.9	6
23	A splice-site variant (c.3289-1G>T) in OTOF underlies profound hearing loss in a Pakistani kindred. BMC Medical Genomics, 2021, 14, 2.	0.7	3
24	Anesthesia, sex and miscarriage history may influence the association between cesarean delivery and autism spectrum disorder. BMC Pediatrics, 2021, 21, 62.	0.7	6
25	Development and validation of a PD-L1/PD-1/CD8 axis-based classifier to predict cancer survival of upper tract urothelial carcinoma after radical nephroureterectomy. Cancer Immunology, Immunotherapy, 2021, 70, 2657-2668.	2.0	4
26	Singleâ€cell analysis of nonhuman primate preimplantation development in comparison to humans and mice. Developmental Dynamics, 2021, 250, 974-985.	0.8	9
27	Cross-Disorder Analysis of De Novo Variants Increases the Power of Prioritising Candidate Genes. Life, 2021, 11, 233.	1.1	0
28	Exploration of the Important Role of Microfibril-Associated Protein 4 Gene in Oral Squamous Cell Carcinoma. Medical Science Monitor, 2021, 27, e931238.	0.5	1
29	Customized de novo mutation detection for any variant calling pipeline: SynthDNM. Bioinformatics, 2021, 37, 3640-3641.	1.8	3
30	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.	1.7	11
31	Extracellular Vesicles from Akkermansia muciniphila Elicit Antitumor Immunity Against Prostate Cancer via Modulation of CD8+ T Cells and Macrophages. International Journal of Nanomedicine, 2021, Volume 16, 2949-2963.	3.3	48
32	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	3.6	50
33	Targeted sequencing and integrative analysis to prioritize candidate genes in neurodevelopmental disorders. Molecular Neurobiology, 2021, 58, 3863-3873.	1.9	5
34	New Model for Estimation of the Age at Onset in Spinocerebellar Ataxia Type 3. Neurology, 2021, 96, e2885-e2895.	1.5	7
35	Harmine targets inhibitor of DNA bindingâ€2 and activator proteinâ€1 to promote preosteoclast PDGFâ€BB production. Journal of Cellular and Molecular Medicine, 2021, 25, 5525-5533.	1.6	6
36	G3BP1 may serve as a potential biomarker of proliferation, apoptosis, and prognosis in oral squamous cell carcinoma. Journal of Oral Pathology and Medicine, 2021, 50, 995-1004.	1.4	13

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37	Mutation pattern and genotype-phenotype correlations of SETD2 in neurodevelopmental disorders. European Journal of Medical Genetics, 2021, 64, 104200.	0.7	5
38	Anxiety and depression in spinocerebellar ataxia patients during the COVID-19 pandemic in China: A cross-sectional study. Journal of Clinical Neuroscience, 2021, 88, 39-46.	0.8	7
39	Evaluation of Peripheral Immune Activation in Amyotrophic Lateral Sclerosis. Frontiers in Neurology, 2021, 12, 628710.	1.1	3
40	Assessment of causal effects of physical activity on neurodegenerative diseases: A Mendelian randomization study. Journal of Sport and Health Science, 2021, 10, 454-461.	3.3	12
41	SLC39A5 dysfunction impairs extracellular matrix synthesis in high myopia pathogenesis. Journal of Cellular and Molecular Medicine, 2021, 25, 8432-8441.	1.6	9
42	Osteocyte exosomes accelerate benign prostatic hyperplasia development. Molecular and Cellular Endocrinology, 2021, 531, 111301.	1.6	5
43	Genetic etiology of a Chinese ataxia cohort: Expanding the mutational spectrum of hereditary ataxias. Parkinsonism and Related Disorders, 2021, 89, 120-127.	1.1	5
44	The autism risk gene CNTN4 modulates dendritic spine formation. Human Molecular Genetics, 2021, 31, 207-218.	1.4	3
45	Joint Analysis of Genome-Wide Association Data Reveals No Genetic Correlations Between Low Back Pain and Neurodegenerative Diseases. Frontiers in Genetics, 2021, 12, 744299.	1.1	4
46	Rare NRXN1 missense variants identified in autism interfered protein degradation and Drosophila sleeping. Journal of Psychiatric Research, 2021, 143, 113-122.	1.5	1
47	A novel defined hypoxia-related gene signature to predict the prognosis of oral squamous cell carcinoma. Annals of Translational Medicine, 2021, 9, 1565-1565.	0.7	11
48	Neuronal Induction of Boneâ€Fat Imbalance through Osteocyte Neuropeptide Y. Advanced Science, 2021, 8, e2100808.	5.6	34
49	Polyglutamine-expanded ataxin3 alter specific gene expressions through changing DNA methylation status in SCA3/MJD. Aging, 2021, 13, 3680-3698.	1.4	4
50	Matrix Metalloproteinases in Relation to Bone Mineral Density: A Two-Sample Mendelian Randomization Study. Frontiers in Genetics, 2021, 12, 754795.	1.1	4
51	Integrative Multiâ˜Omics Analysis Reveals Candidate Biomarkers for Oral Squamous Cell Carcinoma. Frontiers in Oncology, 2021, 11, 794146.	1.3	7
52	Effect of CAG repeats on the age at onset of patients with spinocerebellar ataxia type 2 in China. Journal of Central South University (Medical Sciences), 2021, 46, 793-799.	0.1	1
53	Gene4Denovo: an integrated database and analytic platform for de novo mutations in humans. Nucleic Acids Research, 2020, 48, D913-D926.	6.5	41
54	Phenotypeâ€toâ€genotype approach reveals headâ€circumferenceâ€associated genes in an autism spectrum disorder cohort. Clinical Genetics, 2020, 97, 338-346.	1.0	29

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55	De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. Genetics in Medicine, 2020, 22, 538-546.	1.1	24
56	Genomeâ€wide association analysis of autism identified multiple loci that have been reported as strong signals for neuropsychiatric disorders. Autism Research, 2020, 13, 382-396.	2.1	16
57	Expansion of GGC repeat in the human-specific NOTCH2NLC gene is associated with essential tremor. Brain, 2020, 143, 222-233.	3.7	139
58	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.	0.8	12
59	Excess of RALGAPB de novo variants in neurodevelopmental disorders. European Journal of Medical Genetics, 2020, 63, 104041.	0.7	2
60	Functional relationships between recessive inherited genes and genes with de novo variants in autism spectrum disorder. Molecular Autism, 2020, 11, 75.	2.6	5
61	Fructose-coated Angstrom silver inhibits osteosarcoma growth and metastasis via promoting ROS-dependent apoptosis through the alteration of glucose metabolism by inhibiting PDK. Theranostics, 2020, 10, 7710-7729.	4.6	37
62	Does Maternal Normal Range Thyroid Function Play a Role in Offspring Birth Weight? Evidence From a Mendelian Randomization Analysis. Frontiers in Endocrinology, 2020, 11, 601956.	1.5	6
63	CD147 promotes proliferation and migration of oral cancer cells by inhibiting junctions between Eâ€cadherin and Î²â€catenin. Journal of Oral Pathology and Medicine, 2020, 49, 1019-1029.	1.4	10
64	Ã…ngstrom-scale silver particleâ€“embedded carbomer gel promotes wound healing by inhibiting bacterial colonization and inflammation. Science Advances, 2020, 6, .	4.7	119
65	Biallelic Intronic <scp>AAGGG</scp> Expansion of <scp>RFC1</scp> is Related to Multiple System Atrophy. Annals of Neurology, 2020, 88, 1132-1143.	2.8	41
66	NCKAP1 Disruptive Variants Lead to a Neurodevelopmental Disorder with Core Features of Autism. American Journal of Human Genetics, 2020, 107, 963-976.	2.6	18
67	Genotype and Phenotype Correlations for TBL1XR1 in Neurodevelopmental Disorders. Journal of Molecular Neuroscience, 2020, 70, 2085-2092.	1.1	9
68	Association Between Vitamins and Amyotrophic Lateral Sclerosis: A Center-Based Survey in Mainland China. Frontiers in Neurology, 2020, 11, 488.	1.1	20
69	Hâ€type blood vessels participate in alveolar bone remodeling during murine tooth extraction healing. Oral Diseases, 2020, 26, 998-1009.	1.5	21
70	Fasting before or after wound injury accelerates wound healing through the activation of pro-angiogenic SMOC1 and SCG2. Theranostics, 2020, 10, 3779-3792.	4.6	44
71	Prevalence of Autism Spectrum Disorder in China: A Nationwide Multi-center Population-based Study Among Children Aged 6 to 12 Years. Neuroscience Bulletin, 2020, 36, 961-971.	1.5	179
72	Coffee consumption is not associated with risk of multiple sclerosis: A Mendelian randomization study. Multiple Sclerosis and Related Disorders, 2020, 44, 102300.	0.9	5

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73	The role of genetics in Parkinsonâ€™s disease: a large cohort study in Chinese mainland population. Brain, 2020, 143, 2220-2234.	3.7	97
74	Study on the expression and function of smad family member 7 in oral submucous fibrosis and oral squamous cell carcinoma. Archives of Oral Biology, 2020, 112, 104687.	0.8	5
75	The repertoire of tumor-infiltrating lymphocytes within the microenvironment of oral squamous cell carcinoma reveals immune dysfunction. Cancer Immunology, Immunotherapy, 2020, 69, 465-476.	2.0	32
76	Genetic evidence of gender difference in autism spectrum disorder supports the female-protective effect. Translational Psychiatry, 2020, 10, 4.	2.4	84
77	A founder RDH5 splice site mutation leads to retinitis punctata albescens in two inbred Pakistani kindreds. Ophthalmic Genetics, 2020, 41, 7-12.	0.5	0
78	Pathogenic variants in TNRC6B cause a genetic disorder characterised by developmental delay/intellectual disability and a spectrum of neurobehavioural phenotypes including autism and ADHD. Journal of Medical Genetics, 2020, 57, 717-724.	1.5	14
79	Development of Chinese genetic reference panel for Fragile X Syndrome and its application to the screen of 10,000 Chinese pregnant women and women planning pregnancy. Molecular Genetics & Genomic Medicine, 2020, 8, e1236.	0.6	7
80	Effects of Sleep Disturbances on Behavioral Problems in Preschool Children With Autism Spectrum Disorder. Frontiers in Psychiatry, 2020, 11, 559694.	1.3	7
81	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	5.8	105
82	DEC1: a potential biomarker of malignant transformation in oral leukoplakia. Brazilian Oral Research, 2020, 34, e052.	0.6	6
83	A comparative study of the genetic components of three subcategories of autism spectrum disorder. Molecular Psychiatry, 2019, 24, 1720-1731.	4.1	22
84	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	5.8	150
85	POGZ de novo missense variants in neuropsychiatric disorders. Molecular Genetics & Genomic Medicine, 2019, 7, e900.	0.6	13
86	RNA Expression Profile and Potential Biomarkers in Patients With Spinocerebellar Ataxia Type 3 From Mainland China. Frontiers in Genetics, 2019, 10, 566.	1.1	4
87	Extracellular vesicles from human urine-derived stem cells prevent osteoporosis by transferring CTHRC1 and OPG. Bone Research, 2019, 7, 18.	5.4	66
88	Novel mutation of <scp>EDA</scp> causes new asymmetrical Xâ€linked hypohidrotic ectodermal dysplasia phenotypes in a female. Journal of Dermatology, 2019, 46, 731-733.	0.6	3
89	Alterations of the Gut Microbiota in Multiple System Atrophy Patients. Frontiers in Neuroscience, 2019, 13, 1102.	1.4	42
90	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	5.8	43

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91	Gene-Related Cerebellar Neurodegeneration in SCA3/MJD: A Case-Controlled Imaging-Genetic Study. Frontiers in Neurology, 2019, 10, 1025.	1.1	21
92	Extracellular vesicles from human umbilical cord blood ameliorate bone loss in senile osteoporotic mice. Metabolism: Clinical and Experimental, 2019, 95, 93-101.	1.5	43
93	Genetic and clinical analyses of spinocerebellar ataxia type 8 in mainland China. Journal of Neurology, 2019, 266, 2979-2986.	1.8	7
94	Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	4.7	35
95	Pathogenic missense mutation pattern of forkhead box genes in neurodevelopmental disorders. Molecular Genetics & Genomic Medicine, 2019, 7, e00789.	0.6	9
96	Silver Ã…ngstromâ€Particles: Ã…ngstromâ€Scale Silver Particles as a Promising Agent for Lowâ€Toxicity Broadâ€Spectrum Potent Anticancer Therapy (Adv. Funct. Mater. 23/2019). Advanced Functional Materials, 2019, 29, 1970154.	7.8	1
97	Identification of a potential exosomal biomarker in spinocerebellar ataxia Type 3/Machadoâ€“Joseph disease. Epigenomics, 2019, 11, 1037-1056.	1.0	23
98	Expansion of Human-Specific GGC Repeat in Neuronal Intranuclear Inclusion Disease-Related Disorders. American Journal of Human Genetics, 2019, 105, 166-176.	2.6	212
99	Rare inherited missense variants of POGZ associate with autism risk and disrupt neuronal development. Journal of Genetics and Genomics, 2019, 46, 247-257.	1.7	17
100	A Comparative Study of Genetic Profiles of Key Oncogenesis-Related Genes between Primary Lesions and Matched Lymph Nodes Metastasis in Lung Cancer. Journal of Cancer, 2019, 10, 1642-1650.	1.2	9
101	Ã…ngstromâ€Scale Silver Particles as a Promising Agent for Lowâ€Toxicity Broadâ€Spectrum Potent Anticancer Therapy. Advanced Functional Materials, 2019, 29, 1808556.	7.8	29
102	Suppression of Akt-mTOR pathway rescued the social behavior in Cntnap2-deficient mice. Scientific Reports, 2019, 9, 3041.	1.6	43
103	A novel variation of SERPINC1 caused deep venous thrombosis in a Chinese family. Medicine (United) Tj ETQq1 1 0.784314 rgBT /Ove	0.4	1
104	A de novo mutation of SMYD1 (p.F272L) is responsible for hypertrophic cardiomyopathy in a Chinese patient. Clinical Chemistry and Laboratory Medicine, 2019, 57, 532-539.	1.4	19
105	Long-read sequencing identified intronic repeat expansions inSAMD12from Chinese pedigrees affected with familial cortical myoclonic tremor with epilepsy. Journal of Medical Genetics, 2019, 56, 265-270.	1.5	82
106	Polymorphisms in DNA methylationâ€“related genes are linked to the phenotype of Machado-Joseph disease. Neurobiology of Aging, 2019, 75, 225.e1-225.e8.	1.5	5
107	Common genetic variants shared among five major psychiatric disorders: a large-scale genome-wide combined analysis. Global Clinical and Translational Research, 2019, , 21-30.	0.4	10
108	Altered Gut Microbiome in Autism Spectrum Disorder: Potential Mechanism and Implications for Clinical Intervention. Global Clinical and Translational Research, 2019, , 45-52.	0.4	6

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109	A turn-on fluorescence assay of alkaline phosphatase activity using a DNAâ€“silver nanocluster probe. New Journal of Chemistry, 2018, 42, 4331-4336.	1.4	17
110	Novel West syndrome candidate genes in a Chinese cohort. CNS Neuroscience and Therapeutics, 2018, 24, 1196-1206.	1.9	60
111	Updated frequency analysis of spinocerebellar ataxia in China. Brain, 2018, 141, e22-e22.	3.7	33
112	Genotypeâ€“phenotype correlation and frequency of distribution in a cohort of Chinese Charcotâ€“Marieâ€“Tooth patients associated with GDAP1 mutations. Journal of Neurology, 2018, 265, 637-646.	1.8	14
113	Identification of rare RTN3 variants in Alzheimerâ€™s disease in Han Chinese. Human Genetics, 2018, 137, 141-150.	1.8	22
114	Association of TNF-Î± rs1799964 and IL-1Î² rs16944 polymorphisms with multiple system atrophy in Chinese Han population. International Journal of Neuroscience, 2018, 128, 761-764.	0.8	16
115	Familial paroxysmal kinesigenic dyskinesia is associated with mutations in the KCNA1 gene. Human Molecular Genetics, 2018, 27, 625-637.	1.4	43
116	Increased Reticulon 3 (RTN3) Leads to Obesity and Hypertriglyceridemia by Interacting With Heat Shock Protein Family A (Hsp70) Member 5 (HSPA5). Circulation, 2018, 138, 1828-1838.	1.6	26
117	Differential expression of organic cation transporter 3 in oral submucous fibrosisâ€“associated buccal squamous cell carcinoma. Oral Surgery, Oral Medicine, Oral Pathology and Oral Radiology, 2018, 126, 48-53.	0.2	4
118	A Novel Alzheimer-Associated SNP in Tmp21 Increases Amyloidogenesis. Molecular Neurobiology, 2018, 55, 1862-1870.	1.9	15
119	Excessive UBE3A dosage impairs retinoic acid signaling and synaptic plasticity in autism spectrum disorders. Cell Research, 2018, 28, 48-68.	5.7	95
120	Screening for SH3TC2, PMP2, and BSCL2 Variants in a Cohort of Chinese Patients with Charcot-Marie-Tooth. Chinese Medical Journal, 2018, 131, 151-155.	0.9	3
121	Inherited and multiple de novo mutations in autism/developmental delay risk genes suggest a multifactorial model. Molecular Autism, 2018, 9, 64.	2.6	114
122	Genotype and phenotype correlations for SHANK3 de novo mutations in neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2018, 176, 2668-2676.	0.7	25
123	Genomic landscapes of Chinese sporadic autism spectrum disorders revealed by whole-genome sequencing. Journal of Genetics and Genomics, 2018, 45, 527-538.	1.7	33
124	Coding mutations inNUS1contribute to Parkinsonâ€™s disease. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 11567-11572.	3.3	78
125	Fluorometric aptamer-based determination of ochratoxin A based on the use of graphene oxide and RNase H-aided amplification. Mikrochimica Acta, 2018, 185, 347.	2.5	39
126	Cerebellar lncRNA Expression Profile Analysis of SCA3/MJD Mice. International Journal of Genomics, 2018, 2018, 1-6.	0.8	5

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127	Investigation on modulation of DNA repair pathways in Chinese MJD patients. Neurobiology of Aging, 2018, 71, 267.e5-267.e6.	1.5	5
128	Birt-Hogg-DubÃ© syndrome in two Chinese families with mutations in the FLCN gene. BMC Medical Genetics, 2018, 19, 14.	2.1	13
129	Screening for distress in patients with primary brain tumor using distress thermometer: a systematic review and meta-analysis. BMC Cancer, 2018, 18, 124.	1.1	31
130	Comparing the benefits of chemoradiotherapy and chemotherapy for resectable stage III A/N2 non-small cell lung cancer: a meta-analysis. World Journal of Surgical Oncology, 2018, 16, 8.	0.8	47
131	A Statistical Framework for Mapping Risk Genes from De Novo Mutations in Whole-Genome-Sequencing Studies. American Journal of Human Genetics, 2018, 102, 1031-1047.	2.6	26
132	PAK2 Haploinsufficiency Results in Synaptic Cytoskeleton Impairment and Autism-Related Behavior. Cell Reports, 2018, 24, 2029-2041.	2.9	64
133	Is the High Frequency of Machado-Joseph Disease in China Due to New Mutational Origins?. Frontiers in Genetics, 2018, 9, 740.	1.1	17
134	Parecoxib inhibits glioblastoma cell proliferation, migration and invasion by up-regulating miRNA-29c. Biology Open, 2017, 6, 311-316.	0.6	14
135	Mutation screening of the PRRT2 gene for benign epilepsy with centrotemporal spikes in Chinese mainland population. International Journal of Neuroscience, 2017, 127, 10-13.	0.8	2
136	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.	1.5	18
137	Genome-wide copy number variation analysis in a Chinese autism spectrum disorder cohort. Scientific Reports, 2017, 7, 44155.	1.6	50
138	The genetic spectrum of familial hypercholesterolemia in the central south region of China. Atherosclerosis, 2017, 258, 84-88.	0.4	22
139	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	9.4	443
140	Ubiquitin-related network underlain by (CAG)n loci modulate age at onset in Machado-Joseph disease. Brain, 2017, 140, e25-e25.	3.7	10
141	Vitamin Dâ€related genes are subjected to significant de novo mutation burdens in autism spectrum disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2017, 174, 568-577.	1.1	20
142	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	7.1	152
143	Novel mutations in ADSL for Adenylosuccinate Lyase Deficiency identified by the combination of Trio-WES and constantly updated guidelines. Scientific Reports, 2017, 7, 1625.	1.6	12
144	Rare GCH1 heterozygous variants contributing to Parkinsonâ€™s disease. Brain, 2017, 140, e41-e41.	3.7	21

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145	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.	1.8	12
146	IGHMBP2 -related clinical and genetic features in a cohort of Chinese Charcotâ€“Marieâ€“Tooth disease type 2 patients. Neuromuscular Disorders, 2017, 27, 193-199.	0.3	14
147	The prognostic value of T Lymphoma Invasion and Metastasis 1 (TIAM1) expression in oral squamous cell carcinoma. Journal of Biochemical and Molecular Toxicology, 2017, 31, e21875.	1.4	3
148	Targeted sequencing and functional analysis reveal brain-size-related genes and their networks in autism spectrum disorders. Molecular Psychiatry, 2017, 22, 1282-1290.	4.1	95
149	A novel NHS mutation causes Nance-Horan Syndrome in a Chinese family. BMC Medical Genetics, 2017, 18, 2.	2.1	12
150	Identification of TDP-43 as an oncogene in melanoma and its function during melanoma pathogenesis. Cancer Biology and Therapy, 2017, 18, 8-15.	1.5	21
151	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.	1.1	25
152	A novel splice-site mutation of WRN (c.IVS28+2T>C) identified in a consanguineous family with Werner Syndrome. Molecular Medicine Reports, 2017, 15, 3735-3738.	1.1	2
153	Long-term follow-up of an Alport syndrome patient with a novel mutation of. International Journal of Clinical and Experimental Pathology, 2017, 10, 8709-8714.	0.5	0
154	A Novel Mutation ofFOXC1(R127L) in an Axenfeldâ€“Rieger Syndrome Family with Glaucoma and Multiple Congenital Heart Diseases. Ophthalmic Genetics, 2016, 37, 1-5.	0.5	23
155	An adaptive immune response driven by mature, antigenâ€experienced <scp>T</scp> and <scp>B</scp> cells within the microenvironment of oral squamous cell carcinoma. International Journal of Cancer, 2016, 138, 2952-2962.	2.3	22
156	Association and geneâ€“gene interactions study of reelin signaling pathway related genes with autism in the <scp>H</scp>an <scp>C</scp>hinese population. Autism Research, 2016, 9, 436-442.	2.1	9
157	Prediction of orthostatic hypotension in multiple system atrophy and Parkinson disease. Scientific Reports, 2016, 6, 21649.	1.6	13
158	AKAP2identified as a novel gene mutated in a Chinese family with adolescent idiopathic scoliosis. Journal of Medical Genetics, 2016, 53, 488-493.	1.5	43
159	Variation of global DNA methylation levels with age and in autistic children. Human Genomics, 2016, 10, 31.	1.4	21
160	Mutation detection in Chinese patients with familial hypercholesterolemia. SpringerPlus, 2016, 5, 2095.	1.2	3
161	Real-time monitoring of DNA methyltransferase activity using a hemimethylated smart probe. Molecular and Cellular Probes, 2016, 30, 185-187.	0.9	5
162	(CAG) _n loci as genetic modifiers of age-at-onset in patients with Machado-Joseph disease from mainland China. Brain, 2016, 139, e41-e41.	3.7	37

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163	Label-free fluorescence turn-on detection of uracil DNA glycosylase activity based on G-quadruplex formation. Talanta, 2016, 160, 449-453.	2.9	16
164	Altered plasma levels of chemokines in autism and their association with social behaviors. Psychiatry Research, 2016, 244, 300-305.	1.7	35
165	Methylation analysis of plasma cell-free DNA for breast cancer early detection using bisulfite next-generation sequencing. Tumor Biology, 2016, 37, 13111-13119.	0.8	38
166	miRâ€222 attenuates cisplatinâ€induced cell death by targeting the <scp>PPP</scp>2R2A/Akt/<scp>mTOR</scp> Axis in bladder cancer cells. Journal of Cellular and Molecular Medicine, 2016, 20, 559-567.	1.6	74
167	MFN2â€related genetic and clinical features in a cohort of Chinese CMT2 patients. Journal of the Peripheral Nervous System, 2016, 21, 38-44.	1.4	17
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169	De novo genic mutations among a Chinese autism spectrum disorder cohort. Nature Communications, 2016, 7, 13316.	5.8	293
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