Xia Wang

List of Publications by Year in descending order

		186209	182361
56	3,138	28	51
papers	citations	h-index	g-index

57	57	57	8325
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	3.3	348
2	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	13.9	205
3	Next generation sequencing-based molecular diagnosis of retinitis pigmentosa: identification of a novel genotype-phenotype correlation and clinical refinements. Human Genetics, 2014, 133, 331-345.	1.8	204
4	Molecular diagnostic experience of whole-exome sequencing in adult patients. Genetics in Medicine, 2016, 18, 678-685.	1.1	186
5	Mutations in NMNAT1 cause Leber congenital amaurosis and identify a new disease pathway for retinal degeneration. Nature Genetics, 2012, 44, 1035-1039.	9.4	177
6	Non-invasive prenatal sequencing for multiple Mendelian monogenic disorders using circulating cell-free fetal DNA. Nature Medicine, 2019, 25, 439-447.	15.2	160
7	Toll-like receptor alterations in myelodysplastic syndrome. Leukemia, 2013, 27, 1832-1840.	3.3	139
8	Comprehensive molecular diagnosis of 179 Leber congenital amaurosis and juvenile retinitis pigmentosa patients by targeted next generation sequencing. Journal of Medical Genetics, 2013, 50, 674-688.	1.5	139
9	The Drosophila melanogaster transcriptome by paired-end RNA sequencing. Genome Research, 2011, 21, 315-324.	2.4	123
10	Clinical exome sequencing for fetuses with ultrasound abnormalities and a suspected Mendelian disorder. Genome Medicine, 2018, 10, 74.	3.6	105
11	Next-Generation Sequencingâ€“Based Molecular Diagnosis of a Chinese Patient Cohort With Autosomal Recessive Retinitis Pigmentosa. , 2013, 54, 4158.		85
12	Comprehensive Molecular Diagnosis of a Large Chinese Leber Congenital Amaurosis Cohort. , 2015, 56, 3642.		82
13	Maternal Urinary Triclosan Concentration in Relation to Maternal and Neonatal Thyroid Hormone Levels: A Prospective Study. Environmental Health Perspectives, 2017, 125, 067017.	2.8	76
14	The next generation of population-based spinal muscular atrophy carrier screening: comprehensive pan-ethnic SMN1 copy-number and sequence variant analysis by massively parallel sequencing. Genetics in Medicine, 2017, 19, 936-944.	1.1	70
15	Phenotypic expansion in <scp>DDX</scp>3X â€“ a common cause of intellectual disability in females. Annals of Clinical and Translational Neurology, 2018, 5, 1277-1285.	1.7	66
16	Mutations in human IFT140 cause non-syndromic retinal degeneration. Human Genetics, 2015, 134, 1069-1078.	1.8	62
17	Whole-exome sequencing identifiesALMS1, IQCB1, CNGA3, andMYO7Amutations in patients with leber congenital amaurosis. Human Mutation, 2011, 32, 1450-1459.	1.1	59
18	De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features. American Journal of Human Genetics, 2018, 103, 154-162.	2.6	56

#	Article	IF	Citations
19	A clinical survey of mosaic single nucleotide variants in disease-causing genes detected by exome sequencing. Genome Medicine, 2019, 11, 48.	3.6	55
20	Efficacy of Postnatal InÂVivo Nonsense Suppression Therapy in a Pax6 Mouse Model of Aniridia. Molecular Therapy - Nucleic Acids, 2017, 7, 417-428.	2.3	45
21	MIPEP recessive variants cause a syndrome of left ventricular non-compaction, hypotonia, and infantile death. Genome Medicine, 2016, 8, 106.	3.6	43
22	Copy number variant and runs of homozygosity detection by microarrays enabled more precise molecular diagnoses in 11,020 clinical exome cases. Genome Medicine, 2019, 11, 30.	3.6	42
23	Comprehensive analysis of patients with Stargardt macular dystrophy reveals new genotypeâ€“phenotype correlations and unexpected diagnostic revisions. Genetics in Medicine, 2015, 17, 262-270.	1.1	41
24	ADIPOR1Is Mutated in Syndromic Retinitis Pigmentosa. Human Mutation, 2016, 37, 246-249.	1.1	41
25	Kynurenic acid downregulates IL-17/1L-23 axis in vitro. Molecular and Cellular Biochemistry, 2017, 431, 55-65.	1.4	41
26	Germline mutations in ABL1 cause an autosomal dominant syndrome characterized by congenital heart defects and skeletal malformations. Nature Genetics, 2017, 49, 613-617.	9.4	40
27	Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders. Genome Medicine, 2017, 9, 73.	3.6	39
28	CNVs cause autosomal recessive genetic diseases with or without involvement of SNV/indels. Genetics in Medicine, 2020, 22, 1633-1641.	1.1	36
29	Prenatal exposure to perfluoroalkyl and polyfluoroalkyl substances and childhood atopic dermatitis: a prospective birth cohort study. Environmental Health, 2018, 17, 8.	1.7	33
30	ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 319-330.	2.6	30
31	Deuterated Covalent Organic Frameworks with Significantly Enhanced Luminescence. Angewandte Chemie - International Edition, 2021, 60, 21250-21255.	7.2	30
32	A mouse model of aniridia reveals the in vivo downstream targets of Pax6 driving iris and ciliary body development in the eye. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 60-67.	1.8	24
33	Nonsense suppression therapies in ocular genetic diseases. Cellular and Molecular Life Sciences, 2015, 72, 1931-1938.	2.4	22
34	Sleep disorders and allergic diseases in Chinese toddlers. Sleep Medicine, 2017, 37, 174-179.	0.8	21
35	Widening of the genetic and clinical spectrum of Lambâ€“Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. Genetics in Medicine, 2020, 22, 524-537.	1.1	21
36	Biallelic loss-of-function P4HTM gene variants cause hypotonia, hypoventilation, intellectual disability, dysautonomia, epilepsy, and eye abnormalities (HIDEA syndrome). Genetics in Medicine, 2019, 21, 2355-2363.	1.1	19

#	Article	IF	Citations
37	Maternal prenatal urinary bisphenol A level and child cardio-metabolic risk factors: A prospective cohort study. Environmental Pollution, 2020, 265, 115008.	3.7	18
38	Maternal urinary bisphenol A concentration and thyroid hormone levels of Chinese mothers and newborns by maternal body mass index. Environmental Science and Pollution Research, 2020, 27, 10939-10949.	2.7	17
39	Efficient capture of Sr²⁺ from acidic aqueous solution by an 18-crown-6-ether-based metal organic framework. CrystEngComm, 2021, 23, 3349-3355.	1.3	16
40	Union Makes Strength: A Worldwide Collaborative Genetic and Clinical Study to Provide a Comprehensive Survey of RD3 Mutations and Delineate the Associated Phenotype. PLoS ONE, 2013, 8, e51622.	1.1	16
41	Dawn of ocular gene therapy: implications for molecular diagnosis in retinal disease. Science China Life Sciences, 2013, 56, 125-133.	2.3	15
42	Association Between Down-Regulation of EZH2 and Abnormal Karyotype, Response to Hypomethylation Treatment, and Patient Survival in Myelodysplastic Syndromes. Blood, 2014, 124, 3241-3241.	0.6	15
43	Recurrent mosaic MTOR c.5930C > T (p.Thr1977Ile) variant causing megalencephaly, asymmetric polymicrogyria, and cutaneous pigmentary mosaicism: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2019, 179, 475-479.	0.7	11
44	Application of Next-Generation Sequencing for Genetic Diagnosis in Neonatal Intensive Care Units: Results of a Multicenter Study in China. Frontiers in Genetics, 2020, 11, 565078.	1.1	11
45	X-Linked Retinoschisis: Phenotypic Variability in a Chinese Family. Scientific Reports, 2016, 6, 20118.	1.6	10
46	The expanding clinical phenotype of germline ABL1 â€associated congenital heart defects and skeletal malformations syndrome. Human Mutation, 2020, 41, 1738-1744.	1.1	10
47	Retinal Diseases Caused by Mutations in Genes Not Specifically Associated with the Clinical Diagnosis. PLoS ONE, 2016, 11, e0165405.	1.1	9
48	Prospects and modalities for the treatment of genetic ocular anomalies. Human Genetics, 2019, 138, 1019-1026.	1.8	7
49	Single-crystal-to-single-crystal desolvation in a Ti₃₂ nanoring cluster. CrystEngComm, 2018, 20, 7062-7065.	1.3	6
50	The impact of cesarean delivery on infant DNA methylation. BMC Pregnancy and Childbirth, 2021, 21, 265.	0.9	4
51	GRIPT: a novel case-control analysis method for Mendelian disease gene discovery. Genome Biology, 2018, 19, 203.	3.8	3
52	RNA-based therapies in animal models of Leber congenital amaurosis causing blindness. Precision Clinical Medicine, 2020, 3, 113-126.	1.3	1
53	Family-Based Next-Generation Sequencing Analysis. , 2017, , 321-338.		0
54	Deuterated Covalent Organic Frameworks with Significantly Enhanced Luminescence. Angewandte Chemie, 2021, 133, 21420-21425.	1.6	0

#	Article	IF	Citations
55	Assessment Of EZH2 Expression In CD34+ Bone Marrow Progenitor Cells Of Patients Of Myelodysplastic Syndromes (MDS). Blood, 2013, 122, 2805-2805.	0.6	0
56	The Next Generation Sequencing Based Molecular Diagnosis of Visual Diseases. , 2017, , 51-69.		0