

# Xia Wang

## List of Publications by Year in descending order

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Version: 2024-02-01

56  
papers

3,138  
citations

186209

28  
h-index

182361

51  
g-index

57  
all docs

57  
docs citations

57  
times ranked

8325  
citing authors

#	ARTICLE	IF	CITATIONS
1	The impact of cesarean delivery on infant DNA methylation. BMC Pregnancy and Childbirth, 2021, 21, 265.	0.9	4
2	Deuterated Covalent Organic Frameworks with Significantly Enhanced Luminescence. Angewandte Chemie, 2021, 133, 21420-21425.	1.6	0
3	Deuterated Covalent Organic Frameworks with Significantly Enhanced Luminescence. Angewandte Chemie - International Edition, 2021, 60, 21250-21255.	7.2	30
4	Efficient capture of Sr <sup>2+</sup> from acidic aqueous solution by an 18-crown-6-ether-based metal organic framework. CrystEngComm, 2021, 23, 3349-3355.	1.3	16
5	Widening of the genetic and clinical spectrum of Lambdâ€“Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. Genetics in Medicine, 2020, 22, 524-537.	1.1	21
6	RNA-based therapies in animal models of Leber congenital amaurosis causing blindness. Precision Clinical Medicine, 2020, 3, 113-126.	1.3	1
7	The expanding clinical phenotype of germline <i>ABL1</i> associated congenital heart defects and skeletal malformations syndrome. Human Mutation, 2020, 41, 1738-1744.	1.1	10
8	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
9	Maternal prenatal urinary bisphenol A level and child cardio-metabolic risk factors: A prospective cohort study. Environmental Pollution, 2020, 265, 115008.	3.7	18
10	CNVs cause autosomal recessive genetic diseases with or without involvement of SNV/indels. Genetics in Medicine, 2020, 22, 1633-1641.	1.1	36
11	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
12	A clinical survey of mosaic single nucleotide variants in disease-causing genes detected by exome sequencing. Genome Medicine, 2019, 11, 48.	3.6	55
13	Non-invasive prenatal sequencing for multiple Mendelian monogenic disorders using circulating cell-free fetal DNA. Nature Medicine, 2019, 25, 439-447.	15.2	160
14	Prospects and modalities for the treatment of genetic ocular anomalies. Human Genetics, 2019, 138, 1019-1026.	1.8	7
15	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	13.9	205
16	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
17	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
18	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

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19	ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 319-330.	2.6	30
20	GRIPIT: a novel case-control analysis method for Mendelian disease gene discovery. Genome Biology, 2018, 19, 203.	3.8	3
21	Single-crystal-to-single-crystal desolvation in a Ti <sub>32</sub> nanoring cluster. CrystEngComm, 2018, 20, 7062-7065.	1.3	6
22	Clinical exome sequencing for fetuses with ultrasound abnormalities and a suspected Mendelian disorder. Genome Medicine, 2018, 10, 74.	3.6	105
23	Phenotypic expansion in <i>DDX3X</i> – a common cause of intellectual disability in females. Annals of Clinical and Translational Neurology, 2018, 5, 1277-1285.	1.7	66
24	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
25	Prenatal exposure to perfluoroalkyl and polyfluoroalkyl substances and childhood atopic dermatitis: a prospective birth cohort study. Environmental Health, 2018, 17, 8.	1.7	33
26	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
27	Efficacy of Postnatal In Vivo Nonsense Suppression Therapy in a Pax6 Mouse Model of Aniridia. Molecular Therapy - Nucleic Acids, 2017, 7, 417-428.	2.3	45
28	Family-Based Next-Generation Sequencing Analysis. , 2017, , 321-338.		0
29	Sleep disorders and allergic diseases in Chinese toddlers. Sleep Medicine, 2017, 37, 174-179.	0.8	21
30	Kynurenic acid downregulates IL-17/IL-23 axis in vitro. Molecular and Cellular Biochemistry, 2017, 431, 55-65.	1.4	41
31	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
32	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	3.3	348
33	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
34	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
35	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
36	The Next Generation Sequencing Based Molecular Diagnosis of Visual Diseases. , 2017, , 51-69.		0

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37	X-Linked Retinoschisis: Phenotypic Variability in a Chinese Family. <i>Scientific Reports</i> , 2016, 6, 20118.	1.6	10
38	MIPEP recessive variants cause a syndrome of left ventricular non-compaction, hypotonia, and infantile death. <i>Genome Medicine</i> , 2016, 8, 106.	3.6	43
39	<i>ADIPOR1</i> Is Mutated in Syndromic Retinitis Pigmentosa. <i>Human Mutation</i> , 2016, 37, 246-249.	1.1	41
40	Molecular diagnostic experience of whole-exome sequencing in adult patients. <i>Genetics in Medicine</i> , 2016, 18, 678-685.	1.1	186
41	Retinal Diseases Caused by Mutations in Genes Not Specifically Associated with the Clinical Diagnosis. <i>PLoS ONE</i> , 2016, 11, e0165405.	1.1	9
42	Comprehensive Molecular Diagnosis of a Large Chinese Leber Congenital Amaurosis Cohort. , 2015, 56, 3642.		82
43	Comprehensive analysis of patients with Stargardt macular dystrophy reveals new genotype-phenotype correlations and unexpected diagnostic revisions. <i>Genetics in Medicine</i> , 2015, 17, 262-270.	1.1	41
44	Nonsense suppression therapies in ocular genetic diseases. <i>Cellular and Molecular Life Sciences</i> , 2015, 72, 1931-1938.	2.4	22
45	Mutations in human IFT140 cause non-syndromic retinal degeneration. <i>Human Genetics</i> , 2015, 134, 1069-1078.	1.8	62
46	Next generation sequencing-based molecular diagnosis of retinitis pigmentosa: identification of a novel genotype-phenotype correlation and clinical refinements. <i>Human Genetics</i> , 2014, 133, 331-345.	1.8	204
47	Association Between Down-Regulation of EZH2 and Abnormal Karyotype, Response to Hypomethylation Treatment, and Patient Survival in Myelodysplastic Syndromes. <i>Blood</i> , 2014, 124, 3241-3241.	0.6	15
48	Toll-like receptor alterations in myelodysplastic syndrome. <i>Leukemia</i> , 2013, 27, 1832-1840.	3.3	139
49	Dawn of ocular gene therapy: implications for molecular diagnosis in retinal disease. <i>Science China Life Sciences</i> , 2013, 56, 125-133.	2.3	15
50	Comprehensive molecular diagnosis of 179 Leber congenital amaurosis and juvenile retinitis pigmentosa patients by targeted next generation sequencing. <i>Journal of Medical Genetics</i> , 2013, 50, 674-688.	1.5	139
51	Next-Generation Sequencing-Based Molecular Diagnosis of a Chinese Patient Cohort With Autosomal Recessive Retinitis Pigmentosa. , 2013, 54, 4158.		85
52	Union Makes Strength: A Worldwide Collaborative Genetic and Clinical Study to Provide a Comprehensive Survey of RD3 Mutations and Delineate the Associated Phenotype. <i>PLoS ONE</i> , 2013, 8, e51622.	1.1	16
53	Assessment Of EZH2 Expression In CD34+ Bone Marrow Progenitor Cells Of Patients Of Myelodysplastic Syndromes (MDS). <i>Blood</i> , 2013, 122, 2805-2805.	0.6	0
54	Mutations in NMNAT1 cause Leber congenital amaurosis and identify a new disease pathway for retinal degeneration. <i>Nature Genetics</i> , 2012, 44, 1035-1039.	9.4	177

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55	Whole-exome sequencing identifies <i>ALMS1</i> , <i>IQCB1</i> , <i>CNGA3</i> , and <i>MYO7A</i> mutations in patients with leber congenital amaurosis. <i>Human Mutation</i> , 2011, 32, 1450-1459.	1.1	59
56	The <i>Drosophila melanogaster</i> transcriptome by paired-end RNA sequencing. <i>Genome Research</i> , 2011, 21, 315-324.	2.4	123