

Joanna Crawford

List of Publications by Year in descending order

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68
papers

6,051
citations

117453

34
h-index

88477

70
g-index

76
all docs

76
docs citations

76
times ranked

10797
citing authors

#	ARTICLE	IF	CITATIONS
1	Biallelic <i>PI4KA</i> variants cause neurological, intestinal and immunological disease. <i>Brain</i> , 2021, 144, 3597-3610.	3.7	17
2	Genome sequencing in persistently unsolved white matter disorders. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 144-152.	1.7	26
3	Adult-Diagnosed Nonsyndromic Nephronophthisis in Australian Families Caused by Biallelic NPHP4 Variants. <i>American Journal of Kidney Diseases</i> , 2020, 76, 282-287.	2.1	8
4	De Novo Variants in <i>WDR37</i> Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. <i>American Journal of Human Genetics</i> , 2019, 105, 413-424.	2.6	43
5	Precision Medicine Diagnostics for Rare Kidney Disease: Twitter as a Tool in Clinical Genomic Translation. <i>Kidney Medicine</i> , 2019, 1, 315-318.	1.0	4
6	Heterozygous Variants in the Mechanosensitive Ion Channel <i>TMEM63A</i> Result in Transient Hypomyelination during Infancy. <i>American Journal of Human Genetics</i> , 2019, 105, 996-1004.	2.6	52
7	Biallelic variants in <i>LARS2</i> and <i>KARS</i> cause deafness and (ovario)leukodystrophy. <i>Neurology</i> , 2019, 92, e1225-e1237.	1.5	32
8	Genotype-free demultiplexing of pooled single-cell RNA-seq. <i>Genome Biology</i> , 2019, 20, 290.	3.8	55
9	Leukoencephalopathy due to variants in <i>GFPT1</i> associated congenital myasthenic syndrome. <i>Neurology</i> , 2019, 92, e587-e593.	1.5	14
10	Universal Alternative Splicing of Noncoding Exons. <i>Cell Systems</i> , 2018, 6, 245-255.e5.	2.9	110
11	Identification of miR-29b targets using 3-cyanovinylcarbazole containing mimics. <i>Rna</i> , 2018, 24, 597-608.	1.6	5
12	Patient-iPSC-Derived Kidney Organoids Show Functional Validation of a Ciliopathic Renal Phenotype and Reveal Underlying Pathogenetic Mechanisms. <i>American Journal of Human Genetics</i> , 2018, 102, 816-831.	2.6	157
13	X-linked hypomyelination with spondylometaphyseal dysplasia (H-SMD) associated with mutations in <i>AIFM1</i> . <i>Neurogenetics</i> , 2017, 18, 185-194.	0.7	38
14	Long Noncoding RNAs <i>CUPID1</i> and <i>CUPID2</i> Mediate Breast Cancer Risk at 11q13 by Modulating the Response to DNA Damage. <i>American Journal of Human Genetics</i> , 2017, 101, 255-266.	2.6	77
15	A recurrent de novo mutation in <i>TMEM106B</i> causes hypomyelinating leukodystrophy. <i>Brain</i> , 2017, 140, 3105-3111.	3.7	64
16	Intergenic disease-associated regions are abundant in novel transcripts. <i>Genome Biology</i> , 2017, 18, 241.	3.8	45
17	Whole exome sequencing in patients with white matter abnormalities. <i>Annals of Neurology</i> , 2016, 79, 1031-1037.	2.8	116
18	The <i>Evx1/Evx1as</i> gene locus regulates anterior-posterior patterning during gastrulation. <i>Scientific Reports</i> , 2016, 6, 26657.	1.6	24

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19	Improved definition of the mouse transcriptome via targeted RNA sequencing. <i>Genome Research</i> , 2016, 26, 705-716.	2.4	33
20	Expanding the genotypic spectrum of <i>CCBE1</i> mutations in Hennekam syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2694-2697.	0.7	7
21	Genome-wide discovery of human splicing branchpoints. <i>Genome Research</i> , 2015, 25, 290-303.	2.4	222
22	A novel X-linked trichothiodystrophy associated with a nonsense mutation in <i>RNF113A</i> . <i>Journal of Medical Genetics</i> , 2015, 52, 269-274.	1.5	302
23	Quantitative gene profiling of long noncoding RNAs with targeted RNA sequencing. <i>Nature Methods</i> , 2015, 12, 339-342.	9.0	155
24	Loss-of-Function Alanyl-tRNA Synthetase Mutations Cause an Autosomal-Recessive Early-Onset Epileptic Encephalopathy with Persistent Myelination Defect. <i>American Journal of Human Genetics</i> , 2015, 96, 675-681.	2.6	84
25	Mutations in the voltage-gated potassium channel gene <i>KCNH1</i> cause Temple-Baraitser syndrome and epilepsy. <i>Nature Genetics</i> , 2015, 47, 73-77.	9.4	130
26	Targeted sequencing for gene discovery and quantification using RNA CaptureSeq. <i>Nature Protocols</i> , 2014, 9, 989-1009.	5.5	171
27	Mutations in <i>DARS</i> Cause Hypomyelination with Brain Stem and Spinal Cord Involvement and Leg Spasticity. <i>American Journal of Human Genetics</i> , 2013, 92, 774-780.	2.6	151
28	A De Novo Mutation in the β -Tubulin Gene <i>TUBB4A</i> Results in the Leukoencephalopathy Hypomyelination with Atrophy of the Basal Ganglia and Cerebellum. <i>American Journal of Human Genetics</i> , 2013, 92, 767-773.	2.6	174
29	<i>Babesia divergens</i> and <i>Neospora caninum</i> apical membrane antigen 1 structures reveal selectivity and plasticity in apicomplexan parasite host cell invasion. <i>Protein Science</i> , 2013, 22, 114-127.	3.1	35
30	Targeted RNA sequencing reveals the deep complexity of the human transcriptome. <i>Nature Biotechnology</i> , 2012, 30, 99-104.	9.4	437
31	Expression and Function of the Protein Tyrosine Phosphatase Receptor J (PTPRJ) in Normal Mammary Epithelial Cells and Breast Tumors. <i>PLoS ONE</i> , 2012, 7, e40742.	1.1	22
32	The Melanoma-Upregulated Long Noncoding RNA <i>SPRY4-IT1</i> Modulates Apoptosis and Invasion. <i>Cancer Research</i> , 2011, 71, 3852-3862.	0.4	432
33	Host Cell Invasion by Apicomplexan Parasites: Insights from the Co-Structure of AMA1 with a RON2 Peptide. <i>Science</i> , 2011, 333, 463-467.	6.0	168
34	The Human Mitochondrial Transcriptome. <i>Cell</i> , 2011, 146, 645-658.	13.5	716
35	SNORD-host RNA <i>Zfas1</i> is a regulator of mammary development and a potential marker for breast cancer. <i>Rna</i> , 2011, 17, 878-891.	1.6	321
36	Apicomplexan parasite adhesins: novel strategies for targeting host cell carbohydrates. <i>Current Opinion in Structural Biology</i> , 2010, 20, 551-559.	2.6	15

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37	Structure of the micronemal protein 2 A/I domain from <i>Toxoplasma gondii</i> . <i>Protein Science</i> , 2010, 19, 1985-1990.	3.1	15
38	Structural Characterization of Apical Membrane Antigen 1 (AMA1) from <i>Toxoplasma gondii</i> . <i>Journal of Biological Chemistry</i> , 2010, 285, 15644-15652.	1.6	46
39	Structural and Functional Characterization of SporoSAG. <i>Journal of Biological Chemistry</i> , 2010, 285, 12063-12070.	1.6	37
40	Structural Characterization of the Bradyzoite Surface Antigen (BSR4) from <i>Toxoplasma gondii</i> , a Unique Addition to the Surface Antigen Glycoprotein 1-related Superfamily. <i>Journal of Biological Chemistry</i> , 2009, 284, 9192-9198.	1.6	31
41	Mutation screening in Borjeson-Forssman-Lehmann syndrome: identification of a novel de novo PHF6 mutation in a female patient. <i>Journal of Medical Genetics</i> , 2005, 43, 238-243.	1.5	43
42	FBXO31 Is the Chromosome 16q24.3 Senescence Gene, a Candidate Breast Tumor Suppressor, and a Component of an SCF Complex. <i>Cancer Research</i> , 2005, 65, 11304-11313.	0.4	72
43	1024C>T (R342X) is a recurrent PHF6 mutation also found in the original Borjeson-Forssman-Lehmann syndrome family. <i>European Journal of Human Genetics</i> , 2004, 12, 787-789.	1.4	24
44	The de novo chromosome 16 translocations of two patients with abnormal phenotypes (mental retardation, overlock, and Tf 504). <i>Journal of Medical Genetics</i> , 2002, 39, 111-114.	1.1	148
45	Sequencing, Transcript Identification, and Quantitative Gene Expression Profiling in the Breast Cancer Loss of Heterozygosity Region 16q24.3 Reveal Three Potential Tumor-Suppressor Genes. <i>Genomics</i> , 2002, 80, 303-310.	1.3	42
46	Defining regions of loss of heterozygosity of 16q in breast cancer cell lines. <i>Cancer Genetics and Cytogenetics</i> , 2002, 133, 76-82.	1.0	22
47	Is there a relationship between Wolfram syndrome carrier status and suicide?. <i>American Journal of Medical Genetics Part A</i> , 2002, 114, 343-346.	2.4	30
48	Location and structure of the human FHR-5 gene. <i>Genetica</i> , 2002, 114, 157-161.	0.5	12
49	Isolation, Tissue Distribution, and Chromosomal Localization of a Novel Testis-Specific Human Four-Transmembrane Gene Related to CD20 and FcγRII ² . <i>Biochemical and Biophysical Research Communications</i> , 2001, 280, 374-379.	1.0	15
50	No evidence for association of 5-HT _{2A} receptor polymorphism with suicide. <i>American Journal of Medical Genetics Part A</i> , 2000, 96, 879-880.	2.4	22
51	Y-receptor-like genes GPR72 and GPR73: molecular cloning, genomic organisation and assignment to human chromosome 11q21.1 and 2p14 and mouse chromosome 9 and 6. <i>Biochimica Et Biophysica Acta Gene Regulatory Mechanisms</i> , 2000, 1491, 369-375.	2.4	34
52	Molecular cloning and characterisation of GPR74 a novel G-protein coupled receptor closest related to the Y-receptor family. <i>Molecular Brain Research</i> , 2000, 77, 199-208.	2.5	31
53	Cloning, Characterization, and Chromosomal Location of a Novel Human K ⁺ -Cl ⁻ Cotransporter. <i>Journal of Biological Chemistry</i> , 1999, 274, 10661-10667.	1.6	163
54	Mutation analysis of the Fanconi anaemia A gene in breast tumours with loss of heterozygosity at 16q24.3. <i>British Journal of Cancer</i> , 1999, 79, 1049-1052.	2.9	15

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55	Human HPA endoglycosidase heparanase. Map position 4q21.3. <i>Chromosome Research</i> , 1999, 7, 319-319.	1.0	19
56	Human GALR1 galanin receptor (GALNR1). Map position 18q23. <i>Chromosome Research</i> , 1999, 7, 243-243.	1.0	1
57	Genomic structure and expression analysis of the spastic paraplegia gene, SPG7. <i>Human Genetics</i> , 1999, 105, 139-144.	1.8	15
58	GPR56, a Novel Secretin-like Human G-Protein-Coupled Receptor Gene. <i>Genomics</i> , 1999, 55, 296-305.	1.3	74
59	ThePISSLREGene: Structure, Exon Skipping, and Exclusion as Tumor Suppressor in Breast Cancer. <i>Genomics</i> , 1999, 56, 90-97.	1.3	39
60	Characterization of Copine VII, a New Member of the Copine Family, and Its Exclusion as a Candidate in Sporadic Breast Cancers with Loss of Heterozygosity at 16q24.3. <i>Genomics</i> , 1999, 61, 219-226.	1.3	20
61	Molecular Cloning and Chromosomal Mapping of the Human Homologue of MYB Binding Protein (P160) 1A (MYBBP1A) to 17p13.3. <i>Genomics</i> , 1999, 62, 483-489.	1.3	18
62	Analysis of Australian Crohn's disease pedigrees refines the localization for susceptibility to inflammatory bowel disease on chromosome 16. <i>Annals of Human Genetics</i> , 1998, 62, 291-298.	0.3	102
63	Localization of Human Cadherin Genes to Chromosome Regions Exhibiting Cancer-Related Loss of Heterozygosity. <i>Genomics</i> , 1998, 49, 467-471.	1.3	70
64	Construction of a High-Resolution Physical and Transcription Map of Chromosome 16q24.3: A Region of Frequent Loss of Heterozygosity in Sporadic Breast Cancer. <i>Genomics</i> , 1998, 50, 1-8.	1.3	28
65	Characterization and Screening for Mutations of the Growth Arrest-Specific 11 (GAS11) and C16orf3 Genes at 16q24.3 in Breast Cancer. <i>Genomics</i> , 1998, 52, 325-331.	1.3	47
66	Dynamic mutation loci: allele distributions in different populations. <i>Annals of Human Genetics</i> , 1996, 60, 391-400.	0.3	22
67	Positional cloning of the Fanconi anaemia group A gene. <i>Nature Genetics</i> , 1996, 14, 324-328.	9.4	294
68	A splicing mutation (1898 + 1Gâ†'T) in the CFTR gene causing cystic fibrosis. <i>Human Mutation</i> , 1995, 5, 101-102.	1.1	16