Joanna Crawford

List of Publications by Year
in descending order

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68	6,051	117453	88477
papers	citations	34 h-index	70 g-index

76	76	76	10797
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The Human Mitochondrial Transcriptome. Cell, 2011, 146, 645-658.	13.5	716
2	Targeted RNA sequencing reveals the deep complexity of the human transcriptome. Nature Biotechnology, 2012, 30, 99-104.	9.4	437
3	The Melanoma-Upregulated Long Noncoding RNA SPRY4-IT1 Modulates Apoptosis and Invasion. Cancer Research, 2011, 71, 3852-3862.	0.4	432
4	SNORD-host RNA Zfas1 is a regulator of mammary development and a potential marker for breast cancer. Rna, 2011, 17, 878-891.	1.6	321
5	A novel X-linked trichothiodystrophy associated with a nonsense mutation in RNF113A. Journal of Medical Genetics, 2015, 52, 269-274.	1.5	302
6	Positional cloning of the Fanconi anaemia group A gene. Nature Genetics, 1996, 14, 324-328.	9.4	294
7	Genome-wide discovery of human splicing branchpoints. Genome Research, 2015, 25, 290-303.	2.4	222
8	A De Novo Mutation in the Î²-Tubulin Gene TUBB4A Results in the Leukoencephalopathy Hypomyelination with Atrophy of the Basal Ganglia and Cerebellum. American Journal of Human Genetics, 2013, 92, 767-773.	2.6	174
9	Targeted sequencing for gene discovery and quantification using RNA CaptureSeq. Nature Protocols, 2014, 9, 989-1009.	5.5	171
10	Host Cell Invasion by Apicomplexan Parasites: Insights from the Co-Structure of AMA1 with a RON2 Peptide. Science, 2011, 333, 463-467.	6.0	168
11	Cloning, Characterization, and Chromosomal Location of a Novel Human K+-Clâ^' Cotransporter. Journal of Biological Chemistry, 1999, 274, 10661-10667.	1.6	163
12	Patient-iPSC-Derived Kidney Organoids Show Functional Validation of a Ciliopathic Renal Phenotype and Reveal Underlying Pathogenetic Mechanisms. American Journal of Human Genetics, 2018, 102, 816-831.	2.6	157
13	Quantitative gene profiling of long noncoding RNAs with targeted RNA sequencing. Nature Methods, 2015, 12, 339-342.	9.0	155
14	Mutations in DARS Cause Hypomyelination with Brain Stem and Spinal Cord Involvement and Leg Spasticity. American Journal of Human Genetics, 2013, 92, 774-780.	2.6	151
15	The de novo chromosome 16 translocations of two patients with abnormal phenotypes (mental) Tj ETQq1 1 0.784314 rgBT /Overlock	1.1	140
16	Mutations in the voltage-gated potassium channel gene KCNH1 cause Temple-Baraitser syndrome and epilepsy. Nature Genetics, 2015, 47, 73-77.	9.4	130
17	Whole exome sequencing in patients with white matter abnormalities. Annals of Neurology, 2016, 79, 1031-1037.	2.8	116
18	Universal Alternative Splicing of Noncoding Exons. Cell Systems, 2018, 6, 245-255.e5.	2.9	110

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19	Analysis of Australian Crohn's disease pedigrees refines the localization for susceptibility to inflammatory bowel disease on chromosome 16. Annals of Human Genetics, 1998, 62, 291-298.	0.3	102
20	Loss-of-Function Alanyl-tRNA Synthetase Mutations Cause an Autosomal-Recessive Early-Onset Epileptic Encephalopathy with Persistent Myelination Defect. American Journal of Human Genetics, 2015, 96, 675-681.	2.6	84
21	Long Noncoding RNAs CUPID1 and CUPID2 Mediate Breast Cancer Risk at 11q13 by Modulating the Response to DNA Damage. American Journal of Human Genetics, 2017, 101, 255-266.	2.6	77
22	GPR56, a Novel Secretin-like Human G-Protein-Coupled Receptor Gene. Genomics, 1999, 55, 296-305.	1.3	74
23	FBXO31 Is the Chromosome 16q24.3 Senescence Gene, a Candidate Breast Tumor Suppressor, and a Component of an SCF Complex. Cancer Research, 2005, 65, 11304-11313.	0.4	72
24	Localization of Human Cadherin Genes to Chromosome Regions Exhibiting Cancer-Related Loss of Heterozygosity. Genomics, 1998, 49, 467-471.	1.3	70
25	A recurrent de novo mutation in TMEM106B causes hypomyelinating leukodystrophy. Brain, 2017, 140, 3105-3111.	3.7	64
26	Genotype-free demultiplexing of pooled single-cell RNA-seq. Genome Biology, 2019, 20, 290.	3.8	55
27	Heterozygous Variants in the Mechanosensitive Ion Channel TMEM63A Result in Transient Hypomyelination during Infancy. American Journal of Human Genetics, 2019, 105, 996-1004.	2.6	52
28	Characterization and Screening for Mutations of the Growth Arrest-Specific 11 (GAS11) andC16orf3Genes at 16q24.3 in Breast Cancer. Genomics, 1998, 52, 325-331.	1.3	47
29	Structural Characterization of Apical Membrane Antigen 1 (AMA1) from Toxoplasma gondii. Journal of Biological Chemistry, 2010, 285, 15644-15652.	1.6	46
30	Intergenic disease-associated regions are abundant in novel transcripts. Genome Biology, 2017, 18, 241.	3.8	45
31	Mutation screening in Borjeson-Forssman-Lehmann syndrome: identification of a novel de novo PHF6 mutation in a female patient. Journal of Medical Genetics, 2005, 43, 238-243.	1.5	43
32	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. American Journal of Human Genetics, 2019, 105, 413-424.	2.6	43
33	Sequencing, Transcript Identification, and Quantitative Gene Expression Profiling in the Breast Cancer Loss of Heterozygosity Region 16q24.3 Reveal Three Potential Tumor-Suppressor Genes. Genomics, 2002, 80, 303-310.	1.3	42
34	ThePISSLREGene: Structure, Exon Skipping, and Exclusion as Tumor Suppressor in Breast Cancer. Genomics, 1999, 56, 90-97.	1.3	39
35	X-linked hypomyelination with spondylometaphyseal dysplasia (H-SMD) associated with mutations in AIFM1. Neurogenetics, 2017, 18, 185-194.	0.7	38
36	Structural and Functional Characterization of SporoSAG. Journal of Biological Chemistry, 2010, 285, 12063-12070.	1.6	37

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37	Babesia divergensandNeospora caninumapical membrane antigen 1 structures reveal selectivity and plasticity in apicomplexan parasite host cell invasion. Protein Science, 2013, 22, 114-127.	3.1	35
38	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. Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 2000, 1491, 369-375.	2.4	34
39	Improved definition of the mouse transcriptome via targeted RNA sequencing. Genome Research, 2016, 26, 705-716.	2.4	33
40	Biallelic variants in LARS2 and KARS cause deafness and (ovario)leukodystrophy. Neurology, 2019, 92, e1225-e1237.	1.5	32
41	Molecular cloning and characterisation of GPR74 a novel G-protein coupled receptor closest related to the Y-receptor family. Molecular Brain Research, 2000, 77, 199-208.	2.5	31
42	Structural Characterization of the Bradyzoite Surface Antigen (BSR4) from Toxoplasma gondii, a Unique Addition to the Surface Antigen Glycoprotein 1-related Superfamily. Journal of Biological Chemistry, 2009, 284, 9192-9198.	1.6	31
43	Is there a relationship between Wolfram syndrome carrier status and suicide?. American Journal of Medical Genetics Part A, 2002, 114, 343-346.	2.4	30
44	Construction of a High-Resolution Physical and Transcription Map of Chromosome 16q24.3: A Region of Frequent Loss of Heterozygosity in Sporadic Breast Cancer. Genomics, 1998, 50, 1-8.	1.3	28
45	Genome sequencing in persistently unsolved white matter disorders. Annals of Clinical and Translational Neurology, 2020, 7, 144-152.	1.7	26
46	1024C>T (R342X) is a recurrent PHF6 mutation also found in the original BÃ¶rjesonâ€“Forssmanâ€“Lehmann syndrome family. European Journal of Human Genetics, 2004, 12, 787-789.	1.4	24
47	The Evx1/Evx1as gene locus regulates anterior-posterior patterning during gastrulation. Scientific Reports, 2016, 6, 26657.	1.6	24
48	Dynamic mutation loci: allele distributions in different populations. Annals of Human Genetics, 1996, 60, 391-400.	0.3	22
49	No evidence for association of 5-HT2A receptor polymorphism with suicide. American Journal of Medical Genetics Part A, 2000, 96, 879-880.	2.4	22
50	Defining regions of loss of heterozygosity of 16q in breast cancer cell lines. Cancer Genetics and Cytogenetics, 2002, 133, 76-82.	1.0	22
51	Expression and Function of the Protein Tyrosine Phosphatase Receptor J (PTPRJ) in Normal Mammary Epithelial Cells and Breast Tumors. PLoS ONE, 2012, 7, e40742.	1.1	22
52	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. Genomics, 1999, 61, 219-226.	1.3	20
53	Human HPA endoglycosidase heparanase. Map position 4q21.3. Chromosome Research, 1999, 7, 319-319.	1.0	19
54	Molecular Cloning and Chromosomal Mapping of the Human Homologue of MYB Binding Protein (P160) 1A (MYBBP1A) to 17p13.3. Genomics, 1999, 62, 483-489.	1.3	18

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55	Biallelic PI4KA variants cause neurological, intestinal and immunological disease. Brain, 2021, 144, 3597-3610.	3.7	17
56	A splicing mutation (1898 + 1Gâ†'T) in the CFTR gene causing cystic fibrosis. Human Mutation, 1995, 5, 101-102.	1.1	16
57	Mutation analysis of the Fanconi anaemia A gene in breast tumours with loss of heterozygosity at 16q24.3. British Journal of Cancer, 1999, 79, 1049-1052.	2.9	15
58	Genomic structure and expression analysis of the spastic paraplegia gene, SPG7. Human Genetics, 1999, 105, 139-144.	1.8	15
59	Isolation, Tissue Distribution, and Chromosomal Localization of a Novel Testis-Specific Human Four-Transmembrane Gene Related to CD20 and FcÏµRI-Î². Biochemical and Biophysical Research Communications, 2001, 280, 374-379.	1.0	15
60	Apicomplexan parasite adhesins: novel strategies for targeting host cell carbohydrates. Current Opinion in Structural Biology, 2010, 20, 551-559.	2.6	15
61	Structure of the micronemal protein 2 A/I domain from Toxoplasma gondii. Protein Science, 2010, 19, 1985-1990.	3.1	15
62	Leukoencephalopathy due to variants in GFPT1- associated congenital myasthenic syndrome. Neurology, 2019, 92, e587-e593.	1.5	14
63	Location and structure of the human FHR-5 gene. Genetica, 2002, 114, 157-161.	0.5	12
64	Adult-Diagnosed Nonsyndromic Nephronophthisis in Australian Families Caused by Biallelic NPHP4 Variants. American Journal of Kidney Diseases, 2020, 76, 282-287.	2.1	8
65	Expanding the genotypic spectrum of CCBE1 mutations in Hennekam syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2694-2697.	0.7	7
66	Identification of miR-29b targets using 3-cyanovinylcarbazole containing mimics. Rna, 2018, 24, 597-608.	1.6	5
67	Precision Medicine Diagnostics for Rare Kidney Disease: Twitter as a Tool in Clinical Genomic Translation. Kidney Medicine, 2019, 1, 315-318.	1.0	4
68	Human GALR1 galanin receptor (GALNR1). Map position 18q23. Chromosome Research, 1999, 7, 243-243.	1.0	1