

# Deanna M Church

## List of Publications by Year in descending order

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Version: 2024-02-01

52  
papers

62,782  
citations

87888  
38  
h-index

182427  
51  
g-index

54  
all docs

54  
docs citations

54  
times ranked

74869  
citing authors

#	ARTICLE	IF	CITATIONS
1	Initial sequencing and analysis of the human genome. <i>Nature</i> , 2001, 409, 860-921.	27.8	21,074
2	A global reference for human genetic variation. <i>Nature</i> , 2015, 526, 68-74.	27.8	13,998
3	Initial sequencing and comparative analysis of the mouse genome. <i>Nature</i> , 2002, 420, 520-562.	27.8	6,319
4	Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. <i>Nature</i> , 2007, 447, 799-816.	27.8	4,709
5	Molecular basis of myotonic dystrophy: Expansion of a trinucleotide (CTG) repeat at the 3' end of a transcript encoding a protein kinase family member. <i>Cell</i> , 1992, 68, 799-808.	28.9	2,464
6	Consensus Statement: Chromosomal Microarray Is a First-Tier Clinical Diagnostic Test for Individuals with Developmental Disabilities or Congenital Anomalies. <i>American Journal of Human Genetics</i> , 2010, 86, 749-764.	6.2	2,325
7	ClinVar: public archive of relationships among sequence variation and human phenotype. <i>Nucleic Acids Research</i> , 2014, 42, D980-D985.	14.5	2,270
8	Mutations in the transmembrane domain of FGFR3 cause the most common genetic form of dwarfism, achondroplasia. <i>Cell</i> , 1994, 78, 335-342.	28.9	1,218
9	Database resources of the National Center for Biotechnology. <i>Nucleic Acids Research</i> , 2003, 31, 28-33.	14.5	879
10	Evaluation of GRCh38 and de novo haploid genome assemblies demonstrates the enduring quality of the reference assembly. <i>Genome Research</i> , 2017, 27, 849-864.	5.5	728
11	Direct determination of diploid genome sequences. <i>Genome Research</i> , 2017, 27, 757-767.	5.5	728
12	Multi-platform discovery of haplotype-resolved structural variation in human genomes. <i>Nature Communications</i> , 2019, 10, 1784.	12.8	636
13	Modernizing Reference Genome Assemblies. <i>PLoS Biology</i> , 2011, 9, e1001091.	5.6	458
14	Lineage-Specific Biology Revealed by a Finished Genome Assembly of the Mouse. <i>PLoS Biology</i> , 2009, 7, e1000112.	5.6	419
15	An evidence-based approach to establish the functional and clinical significance of copy number variants in intellectual and developmental disabilities. <i>Genetics in Medicine</i> , 2011, 13, 777-784.	2.4	371
16	A genome-wide comparison of recent chimpanzee and human segmental duplications. <i>Nature</i> , 2005, 437, 88-93.	27.8	353
17	Assembly: a resource for assembled genomes at NCBI. <i>Nucleic Acids Research</i> , 2016, 44, D73-D80.	14.5	292
18	Isolation of genes from complex sources of mammalian genomic DNA using exon amplification. <i>Nature Genetics</i> , 1994, 6, 98-105.	21.4	291

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19	Spidey: A Tool for mRNA-to-Genomic Alignments. <i>Genome Research</i> , 2001, 11, 1952-1957.	5.5	285
20	The emergent landscape of the mouse gut endoderm at single-cell resolution. <i>Nature</i> , 2019, 569, 361-367.	27.8	285
21	Structure and expression of the Huntington's disease gene: Evidence against simple inactivation due to an expanded CAG repeat. <i>Somatic Cell and Molecular Genetics</i> , 1994, 20, 27-38.	0.7	246
22	Shotgun sequence assembly and recent segmental duplications within the human genome. <i>Nature</i> , 2004, 431, 927-930.	27.8	228
23	Cross-Species Sequence Comparisons: A Review of Methods and Available Resources. <i>Genome Research</i> , 2003, 13, 1-12.	5.5	210
24	Mouse segmental duplication and copy number variation. <i>Nature Genetics</i> , 2008, 40, 909-914.	21.4	209
25	Database resources of the National Center for Biotechnology Information: 2002 update. <i>Nucleic Acids Research</i> , 2002, 30, 13-16.	14.5	184
26	Resolving the full spectrum of human genome variation using Linked-Reads. <i>Genome Research</i> , 2019, 29, 635-645.	5.5	182
27	Completing the map of human genetic variation. <i>Nature</i> , 2007, 447, 161-165.	27.8	178
28	A general approach for detecting expressed mutations in AML cells using single cell RNA-sequencing. <i>Nature Communications</i> , 2019, 10, 3660.	12.8	147
29	Good laboratory practice for clinical next-generation sequencing informatics pipelines. <i>Nature Biotechnology</i> , 2015, 33, 689-693.	17.5	134
30	Single haplotype assembly of the human genome from a hydatidiform mole. <i>Genome Research</i> , 2014, 24, 2066-2076.	5.5	133
31	Sequence variations in the public human genome data reflect a bottlenecked population history. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2003, 100, 376-381.	7.1	113
32	Analysis of Segmental Duplications and Genome Assembly in the Mouse. <i>Genome Research</i> , 2004, 14, 789-801.	5.5	106
33	A radiation hybrid map of mouse genes. <i>Nature Genetics</i> , 2001, 29, 201-205.	21.4	93
34	Mitochondrial Disease Sequence Data Resource (MSeqDR): A global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. <i>Molecular Genetics and Metabolism</i> , 2015, 114, 388-396.	1.1	76
35	Public data archives for genomic structural variation. <i>Nature Genetics</i> , 2010, 42, 813-814.	21.4	71
36	Haplotyping the <i>Vitis</i> collinear core genome with rhAmpSeq improves marker transferability in a diverse genus. <i>Nature Communications</i> , 2020, 11, 413.	12.8	52

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37	Candidate Single Nucleotide Polymorphism Selection using Publicly Available Tools: A Guide for Epidemiologists. American Journal of Epidemiology, 2006, 164, 794-804.	3.4	49
38	A High-Resolution Physical and Transcript Map of the Cri du Chat Region of Human Chromosome 5p. Genome Research, 1997, 7, 787-801.	5.5	44
39	Mouse Chromosome 2. Mammalian Genome, 1999, 10, 941-941.	2.2	39
40	Identification of human chromosome 9 specific genes using exon amplification. Human Molecular Genetics, 1993, 2, 1915-1920.	2.9	30
41	A gene from chromosome 4p 16.3 with similarity to a superfamily of transporter proteins. Human Molecular Genetics, 1993, 2, 673-676.	2.9	28
42	Principles and Recommendations for Standardizing the Use of the Next-Generation Sequencing Variant File in Clinical Settings. Journal of Molecular Diagnostics, 2017, 19, 417-426.	2.8	19
43	Efficiency and specificity of gene isolation by exon amplification. Mammalian Genome, 1993, 4, 466-474.	2.2	18
44	Connecting Sequence and Biology in the Laboratory Mouse. Genome Research, 2003, 13, 1505-1519.	5.5	18
45	Transcript Mapping of the Human Chromosome 11q12-q13.1 Gene-Rich Region Identifies Several Newly Described Conserved Genes. Genomics, 1998, 49, 419-429.	2.9	16
46	[6] Gene identification by exon amplification. Methods in Enzymology, 1999, 303, 83-99.	1.0	14
47	A Radiation Hybrid Map of Human Chromosome 5 with Integration of Cytogenetic, Genetic, and Transcript Maps. Genome Research, 1997, 7, 897-909.	5.5	11
48	WebACGH. Applied Bioinformatics, 2006, 5, 125-130.	1.6	5
49	Genomes for all. Nature Biotechnology, 2018, 36, 815-816.	17.5	5
50	Exon trapping and sequence-based methods of gene finding in transcript mapping of human 4p 16.3. Somatic Cell and Molecular Genetics, 1997, 23, 413-427.	0.7	4
51	Thousands of human sequences provide deep insight into single genomes. Nature, 2020, 581, 385-386.	27.8	1
52	Accessing the Human Genome. Current Protocols in Human Genetics, 2002, 34, Unit 6.9.	3.5	0