Deborah A Nickerson

List of Publications by Year
in descending order

53	22,589	172457	175258
papers	citations	29 h-index	52 g-index

64	64	64	44629
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	27.8	13,998
2	Targeted capture and massively parallel sequencing of 12 human exomes. Nature, 2009, 461, 272-276.	27.8	1,801
3	Sequencing of 53,831 diverse genomes from the NHLBI TOPMed Program. Nature, 2021, 590, 290-299.	27.8	1,069
4	Analysis of 6,515 exomes reveals the recent origin of most human protein-coding variants. Nature, 2013, 493, 216-220.	27.8	898
5	Optimal Unified Approach for Rare-Variant Association Testing with Application to Small-Sample Case-Control Whole-Exome Sequencing Studies. American Journal of Human Genetics, 2012, 91, 224-237.	6.2	880
6	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215.	6.2	574
7	Copy number variation detection and genotyping from exome sequence data. Genome Research, 2012, 22, 1525-1532.	5.5	550
8	De novo rates and selection of large copy number variation. Genome Research, 2010, 20, 1469-1481.	5.5	264
9	Genetic Ancestry in Lung-Function Predictions. New England Journal of Medicine, 2010, 363, 321-330.	27.0	230
10	Cryptic transmission of SARS-CoV-2 in Washington state. Science, 2020, 370, 571-575.	12.6	217
11	Whole-Exome Sequencing Identifies Rare and Low-Frequency Coding Variants Associated with LDL Cholesterol. American Journal of Human Genetics, 2014, 94, 233-245.	6.2	193
12	Mendelian Gene Discovery: Fast and Furious with No End in Sight. American Journal of Human Genetics, 2019, 105, 448-455.	6.2	166
13	RNF213 Rare Variants in an Ethnically Diverse Population With Moyamoya Disease. Stroke, 2014, 45, 3200-3207.	2.0	129
14	PRIMUS: Rapid Reconstruction of Pedigrees from Genome-wide Estimates of Identity by Descent. American Journal of Human Genetics, 2014, 95, 553-564.	6.2	129
15	Early Detection of Covid-19 through a Citywide Pandemic Surveillance Platform. New England Journal of Medicine, 2020, 383, 185-187.	27.0	97
16	PGRNseq. Pharmacogenetics and Genomics, 2016, 26, 161-168.	1.5	93
17	Stargazer: a software tool for calling star alleles from next-generation sequencing data using CYP2D6 as a model. Genetics in Medicine, 2019, 21, 361-372.	2.4	86
18	GGC Repeat Expansion and Exon 1 Methylation of XYLT1 Is a Common Pathogenic Variant in Baratela-Scott Syndrome. American Journal of Human Genetics, 2019, 104, 35-44.	6.2	81

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19	Mutations in ECEL1 Cause Distal Arthrogryposis Type 5D. American Journal of Human Genetics, 2013, 92, 150-156.	6.2	71
20	Common and rare von Willebrand factor (VWF) coding variants, VWF levels, and factor VIII levels in African Americans: the NHLBI Exome Sequencing Project. Blood, 2013, 122, 590-597.	1.4	70
21	Calling Star Alleles With Stargazer in 28 Pharmacogenes With Whole Genome Sequences. Clinical Pharmacology and Therapeutics, 2019, 106, 1328-1337.	4.7	57
22	Maternal Modifiers and Parent-of-Origin Bias of the Autism-Associated 16p11.2 CNV. American Journal of Human Genetics, 2016, 98, 45-57.	6.2	55
23	Whole-Genome Analysis Reveals that Mutations in Inositol Polyphosphate Phosphatase-like 1 Cause Opsismodysplasia. American Journal of Human Genetics, 2013, 92, 137-143.	6.2	53
24	Large-Scale Exome-wide Association Analysis Identifies Loci for White Blood Cell Traits and Pleiotropy with Immune-Mediated Diseases. American Journal of Human Genetics, 2016, 99, 22-39.	6.2	50
25	Utilizing Graph Theory to Select the Largest Set of Unrelated Individuals for Genetic Analysis. Genetic Epidemiology, 2013, 37, 136-141.	1.3	47
26	Genetic and Nongenetic Factors Associated with Protein Abundance of Flavin-Containing Monooxygenase 3 in Human Liver. Journal of Pharmacology and Experimental Therapeutics, 2017, 363, 265-274.	2.5	43
27	Hepatic Abundance and Activity of Androgen- and Drug-Metabolizing Enzyme UGT2B17 Are Associated with Genotype, Age, and Sex. Drug Metabolism and Disposition, 2018, 46, 888-896.	3.3	42
28	Interrogation of <scp>CYP</scp>2D6 Structural Variant Alleles Improves the Correlation Between <scp>CYP</scp>2D6 Genotype and <scp>CYP</scp>2D6â€Mediated Metabolic Activity. Clinical and Translational Science, 2020, 13, 147-156.	3.1	42
29	Multiplexed Functional Assessment of Genetic Variants in CARD11. American Journal of Human Genetics, 2020, 107, 1029-1043.	6.2	38
30	PADRE: Pedigree-Aware Distant-Relationship Estimation. American Journal of Human Genetics, 2016, 99, 154-162.	6.2	36
31	TCIRG1-Associated Congenital Neutropenia. Human Mutation, 2014, 35, 824-827.	2.5	35
32	The Seattle Flu Study: a multiarm community-based prospective study protocol for assessing influenza prevalence, transmission and genomic epidemiology. BMJ Open, 2020, 10, e037295.	1.9	25
33	Genomic characterization of the RH locus detects complex and novel structural variation in multi-ethnic cohorts. Genetics in Medicine, 2019, 21, 477-486.	2.4	24
34	CYP2C8, CYP2C9, and CYP2C19 Characterization Using Next-Generation Sequencing and Haplotype Analysis. Journal of Molecular Diagnostics, 2022, 24, 337-350.	2.8	23
35	Raceâ€Specific Influence of <scp>CYP</scp>4F2 on Dose and Risk of Hemorrhage Among Warfarin Users. Pharmacotherapy, 2016, 36, 263-272.	2.6	22
36	Mutations in MYLPF Cause a Novel Segmental Amyoplasia that Manifests as Distal Arthrogryposis. American Journal of Human Genetics, 2020, 107, 293-310.	6.2	21

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37	Multigenic Disease and Bilineal Inheritance in Dilated Cardiomyopathy Is Illustrated in Nonsegregating LMNA Pedigrees. Circulation Genomic and Precision Medicine, 2018, 11, e002038.	3.6	20
38	Germline SAMD9L truncation variants trigger global translational repression. Journal of Experimental Medicine, 2021, 218, .	8.5	20
39	Evaluating Specimen Quality and Results from a Community-Wide, Home-Based Respiratory Surveillance Study. Journal of Clinical Microbiology, 2021, 59, .	3.9	17
40	Whole-exome imputation of sequence variants identified two novel alleles associated with adult body height in African Americans. Human Molecular Genetics, 2014, 23, 6607-6615.	2.9	14
41	Pharmacogenomics of Nicotine Metabolism: Novel CYP2A6 and CYP2B6 Genetic Variation Patterns in Alaska Native and American Indian Populations. Nicotine and Tobacco Research, 2020, 22, 910-918.	2.6	14
42	Whole-genome sequencing in diverse subjects identifies genetic correlates of leukocyte traits: The NHLBI TOPMed program. American Journal of Human Genetics, 2021, 108, 1836-1851.	6.2	14
43	Influence of common and rare genetic variation on warfarin dose among Africanâ€“Americans and Europeanâ€“Americans using the exome array. Pharmacogenomics, 2017, 18, 1059-1073.	1.3	12
44	PRIMUS: improving pedigree reconstruction using mitochondrial and Y haplotypes. Bioinformatics, 2016, 32, 596-598.	4.1	11
45	Whole-Genome Sequencing Identifies Novel Functional Loci Associated with Lung Function in Puerto Rican Youth. American Journal of Respiratory and Critical Care Medicine, 2020, 202, 962-972.	5.6	11
46	Opsismodysplasia resulting from an insertion mutation in the SH2 domain, which destabilizes INPPL1. American Journal of Medical Genetics, Part A, 2014, 164, 2407-2411.	1.2	10
47	Whole genome sequence analysis of platelet traits in the NHLBI Trans-Omics for Precision Medicine (TOPMed) initiative. Human Molecular Genetics, 2022, 31, 347-361.	2.9	9
48	Diversity of variant alleles encoding <scp>Kidd, Duffy, and Kell</scp> antigens in individuals with sickle cell disease using whole genome sequencing data from the <scp>NHLBI TOPMed Program</scp>. Transfusion, 2021, 61, 603-616.	1.6	7
49	Analysis of exome sequencing data sets reveals structural variation in the coding region of ABO in individuals of African ancestry. Transfusion, 2016, 56, 2744-2749.	1.6	5
50	Further delineation of van den Endeâ€Gupta syndrome: Genetic heterogeneity and overlap with congenital heart defects and skeletal malformations syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2136-2149.	1.2	5
51	Diagnostic Accuracy of an At-Home, Rapid Self-test for Influenza: Prospective Comparative Accuracy Study. JMIR Public Health and Surveillance, 2022, 8, e28268.	2.6	5
52	Comparable Specimen Collection from Both Ends of At-Home Midturbinate Swabs. Journal of Clinical Microbiology, 2021, 59, .	3.9	2
53	Exome sequencing identifies genetic variants in anophthalmia and microphthalmia. American Journal of Medical Genetics, Part A, 2022, 188, 2376-2388.	1.2	2