

Deborah A Nickerson

List of Publications by Year in descending order

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Version: 2024-02-01

53
papers

22,589
citations

172457

29
h-index

175258

52
g-index

64
all docs

64
docs citations

64
times ranked

44629
citing authors

#	ARTICLE	IF	CITATIONS
1	Whole genome sequence analysis of platelet traits in the NHLBI Trans-Omics for Precision Medicine (TOPMed) initiative. <i>Human Molecular Genetics</i> , 2022, 31, 347-361.	2.9	9
2	Diagnostic Accuracy of an At-Home, Rapid Self-test for Influenza: Prospective Comparative Accuracy Study. <i>JMIR Public Health and Surveillance</i> , 2022, 8, e28268.	2.6	5
3	CYP2C8, CYP2C9, and CYP2C19 Characterization Using Next-Generation Sequencing and Haplotype Analysis. <i>Journal of Molecular Diagnostics</i> , 2022, 24, 337-350.	2.8	23
4	Exome sequencing identifies genetic variants in anophthalmia and microphthalmia. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2376-2388.	1.2	2
5	Diversity of variant alleles encoding Kidd, Duffy, and Kell antigens in individuals with sickle cell disease using whole genome sequencing data from the NHLBI TOPMed Program. <i>Transfusion</i> , 2021, 61, 603-616.	1.6	7
6	Sequencing of 53,831 diverse genomes from the NHLBI TOPMed Program. <i>Nature</i> , 2021, 590, 290-299.	27.8	1,069
7	Further delineation of van den Ende-Gupta syndrome: Genetic heterogeneity and overlap with congenital heart defects and skeletal malformations syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2136-2149.	1.2	5
8	Germline SAMD9L truncation variants trigger global translational repression. <i>Journal of Experimental Medicine</i> , 2021, 218, .	8.5	20
9	Comparable Specimen Collection from Both Ends of At-Home Midturbinate Swabs. <i>Journal of Clinical Microbiology</i> , 2021, 59, .	3.9	2
10	Evaluating Specimen Quality and Results from a Community-Wide, Home-Based Respiratory Surveillance Study. <i>Journal of Clinical Microbiology</i> , 2021, 59, .	3.9	17
11	Whole-genome sequencing in diverse subjects identifies genetic correlates of leukocyte traits: The NHLBI TOPMed program. <i>American Journal of Human Genetics</i> , 2021, 108, 1836-1851.	6.2	14
12	Pharmacogenomics of Nicotine Metabolism: Novel CYP2A6 and CYP2B6 Genetic Variation Patterns in Alaska Native and American Indian Populations. <i>Nicotine and Tobacco Research</i> , 2020, 22, 910-918.	2.6	14
13	Interrogation of CYP2D6 Structural Variant Alleles Improves the Correlation Between CYP2D6 Genotype and CYP2D6-Mediated Metabolic Activity. <i>Clinical and Translational Science</i> , 2020, 13, 147-156.	3.1	42
14	The Seattle Flu Study: a multiarm community-based prospective study protocol for assessing influenza prevalence, transmission and genomic epidemiology. <i>BMJ Open</i> , 2020, 10, e037295.	1.9	25
15	Mutations in MYLPH Cause a Novel Segmental Amyoplasia that Manifests as Distal Arthrogryposis. <i>American Journal of Human Genetics</i> , 2020, 107, 293-310.	6.2	21
16	Cryptic transmission of SARS-CoV-2 in Washington state. <i>Science</i> , 2020, 370, 571-575.	12.6	217
17	Multiplexed Functional Assessment of Genetic Variants in CARD11. <i>American Journal of Human Genetics</i> , 2020, 107, 1029-1043.	6.2	38
18	Whole-Genome Sequencing Identifies Novel Functional Loci Associated with Lung Function in Puerto Rican Youth. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2020, 202, 962-972.	5.6	11

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19	Early Detection of Covid-19 through a Citywide Pandemic Surveillance Platform. <i>New England Journal of Medicine</i> , 2020, 383, 185-187.	27.0	97
20	Stargazer: a software tool for calling star alleles from next-generation sequencing data using CYP2D6 as a model. <i>Genetics in Medicine</i> , 2019, 21, 361-372.	2.4	86
21	Genomic characterization of the RH locus detects complex and novel structural variation in multi-ethnic cohorts. <i>Genetics in Medicine</i> , 2019, 21, 477-486.	2.4	24
22	Mendelian Gene Discovery: Fast and Furious with No End in Sight. <i>American Journal of Human Genetics</i> , 2019, 105, 448-455.	6.2	166
23	Calling Star Alleles With Stargazer in 28 Pharmacogenes With Whole Genome Sequences. <i>Clinical Pharmacology and Therapeutics</i> , 2019, 106, 1328-1337.	4.7	57
24	GCC Repeat Expansion and Exon 1 Methylation of XYL1 Is a Common Pathogenic Variant in Baratela-Scott Syndrome. <i>American Journal of Human Genetics</i> , 2019, 104, 35-44.	6.2	81
25	Hepatic Abundance and Activity of Androgen- and Drug-Metabolizing Enzyme UGT2B17 Are Associated with Genotype, Age, and Sex. <i>Drug Metabolism and Disposition</i> , 2018, 46, 888-896.	3.3	42
26	Multigenic Disease and Bilineal Inheritance in Dilated Cardiomyopathy Is Illustrated in Nonsegregating LMNA Pedigrees. <i>Circulation Genomic and Precision Medicine</i> , 2018, 11, e002038.	3.6	20
27	Genetic and Nongenetic Factors Associated with Protein Abundance of Flavin-Containing Monooxygenase 3 in Human Liver. <i>Journal of Pharmacology and Experimental Therapeutics</i> , 2017, 363, 265-274.	2.5	43
28	Influence of common and rare genetic variation on warfarin dose among African-American and European-American using the exome array. <i>Pharmacogenomics</i> , 2017, 18, 1059-1073.	1.3	12
29	PADRE: Pedigree-Aware Distant-Relationship Estimation. <i>American Journal of Human Genetics</i> , 2016, 99, 154-162.	6.2	36
30	Analysis of exome sequencing data sets reveals structural variation in the coding region of <i>ABO</i> in individuals of African ancestry. <i>Transfusion</i> , 2016, 56, 2744-2749.	1.6	5
31	PGRNseq. <i>Pharmacogenetics and Genomics</i> , 2016, 26, 161-168.	1.5	93
32	Race-Specific Influence of <i>CYP4F2</i> on Dose and Risk of Hemorrhage Among Warfarin Users. <i>Pharmacotherapy</i> , 2016, 36, 263-272.	2.6	22
33	Large-Scale Exome-wide Association Analysis Identifies Loci for White Blood Cell Traits and Pleiotropy with Immune-Mediated Diseases. <i>American Journal of Human Genetics</i> , 2016, 99, 22-39.	6.2	50
34	Maternal Modifiers and Parent-of-Origin Bias of the Autism-Associated 16p11.2 CNV. <i>American Journal of Human Genetics</i> , 2016, 98, 45-57.	6.2	55
35	PRIMUS: improving pedigree reconstruction using mitochondrial and Y haplotypes. <i>Bioinformatics</i> , 2016, 32, 596-598.	4.1	11
36	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. <i>American Journal of Human Genetics</i> , 2015, 97, 199-215.	6.2	574

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37	A global reference for human genetic variation. <i>Nature</i> , 2015, 526, 68-74.	27.8	13,998
38	Whole-exome imputation of sequence variants identified two novel alleles associated with adult body height in African Americans. <i>Human Molecular Genetics</i> , 2014, 23, 6607-6615.	2.9	14
39	<i>RNF213</i> Rare Variants in an Ethnically Diverse Population With Moyamoya Disease. <i>Stroke</i> , 2014, 45, 3200-3207.	2.0	129
40	<i>TCIRG1</i> -Associated Congenital Neutropenia. <i>Human Mutation</i> , 2014, 35, 824-827.	2.5	35
41	Opsismodysplasia resulting from an insertion mutation in the SH2 domain, which destabilizes INPPL1. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2407-2411.	1.2	10
42	Whole-Exome Sequencing Identifies Rare and Low-Frequency Coding Variants Associated with LDL Cholesterol. <i>American Journal of Human Genetics</i> , 2014, 94, 233-245.	6.2	193
43	PRIMUS: Rapid Reconstruction of Pedigrees from Genome-wide Estimates of Identity by Descent. <i>American Journal of Human Genetics</i> , 2014, 95, 553-564.	6.2	129
44	Mutations in <i>ECEL1</i> Cause Distal Arthrogyryposis Type 5D. <i>American Journal of Human Genetics</i> , 2013, 92, 150-156.	6.2	71
45	Analysis of 6,515 exomes reveals the recent origin of most human protein-coding variants. <i>Nature</i> , 2013, 493, 216-220.	27.8	898
46	Whole-Genome Analysis Reveals that Mutations in Inositol Polyphosphate Phosphatase-like 1 Cause Opsismodysplasia. <i>American Journal of Human Genetics</i> , 2013, 92, 137-143.	6.2	53
47	Utilizing Graph Theory to Select the Largest Set of Unrelated Individuals for Genetic Analysis. <i>Genetic Epidemiology</i> , 2013, 37, 136-141.	1.3	47
48	Common and rare von Willebrand factor (VWF) coding variants, VWF levels, and factor VIII levels in African Americans: the NHLBI Exome Sequencing Project. <i>Blood</i> , 2013, 122, 590-597.	1.4	70
49	Copy number variation detection and genotyping from exome sequence data. <i>Genome Research</i> , 2012, 22, 1525-1532.	5.5	550
50	Optimal Unified Approach for Rare-Variant Association Testing with Application to Small-Sample Case-Control Whole-Exome Sequencing Studies. <i>American Journal of Human Genetics</i> , 2012, 91, 224-237.	6.2	880
51	De novo rates and selection of large copy number variation. <i>Genome Research</i> , 2010, 20, 1469-1481.	5.5	264
52	Genetic Ancestry in Lung-Function Predictions. <i>New England Journal of Medicine</i> , 2010, 363, 321-330.	27.0	230
53	Targeted capture and massively parallel sequencing of 12 human exomes. <i>Nature</i> , 2009, 461, 272-276.	27.8	1,801