Anita L Destefano

List of Publications by Year
in descending order

137 papers	22,336 citations	30047 54 h-index	14736 127 g-index
152 all docs	152 docs citations	152 times ranked	27176 citing authors

#	Article	IF	Citations
1	Meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for Alzheimer's disease. Nature Genetics, 2013, 45, 1452-1458.	9.4	3,741
2	Genetic meta-analysis of diagnosed Alzheimerâ€™s disease identifies new risk loci and implicates AÎ², tau, immunity and lipid processing. Nature Genetics, 2019, 51, 414-430.	9.4	1,962
3	Common variants at ABCA7, MS4A6A/MS4A4E, EPHA1, CD33 and CD2AP are associated with Alzheimer's disease. Nature Genetics, 2011, 43, 429-435.	9.4	1,708
4	Large-scale meta-analysis of genome-wide association data identifies six new risk loci for Parkinson's disease. Nature Genetics, 2014, 46, 989-993.	9.4	1,685
5	Multiancestry genome-wide association study of 520,000 subjects identifies 32 loci associated with stroke and stroke subtypes. Nature Genetics, 2018, 50, 524-537.	9.4	1,124
6	Genome-wide Analysis of Genetic Loci Associated With Alzheimer Disease. JAMA - Journal of the American Medical Association, 2010, 303, 1832.	3.8	1,064
7	Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. Nature Genetics, 2017, 49, 1373-1384.	9.4	783
8	Evidence for a Gene Influencing Blood Pressure on Chromosome 17. Hypertension, 2000, 36, 477-483.	1.3	534
9	Comprehensive Research Synopsis and Systematic Meta-Analyses in Parkinson's Disease Genetics: The PDGene Database. PLoS Genetics, 2012, 8, e1002548.	1.5	495
10	Genetic risk factors for ischaemic stroke and its subtypes (the METASTROKE Collaboration): a meta-analysis of genome-wide association studies. Lancet Neurology, The, 2012, 11, 951-962.	4.9	445
11	Genomewide Association Studies of Stroke. New England Journal of Medicine, 2009, 360, 1718-1728.	13.9	420
12	Genomewide association study for susceptibility genes contributing to familial Parkinson disease. Human Genetics, 2009, 124, 593-605.	1.8	410
13	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. Nature Genetics, 2011, 43, 1005-1011.	9.4	403
14	A common haplotype lowers PU.1 expression in myeloid cells and delays onset of Alzheimer's disease. Nature Neuroscience, 2017, 20, 1052-1061.	7.1	330
15	Shared Genetic Susceptibility to Ischemic Stroke and Coronary Artery Disease. Stroke, 2014, 45, 24-36.	1.0	302
16	Metaâ€analysis of Parkinson's Disease: Identification of a novel locus, RIT2. Annals of Neurology, 2012, 71, 370-384.	2.8	264
17	Novel genetic loci associated with hippocampal volume. Nature Communications, 2017, 8, 13624.	5.8	250
18	Serum Brain-Derived Neurotrophic Factor and the Risk for Dementia. JAMA Neurology, 2014, 71, 55.	4.5	219

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19	Novel genetic loci underlying human intracranial volume identified through genome-wide association. Nature Neuroscience, 2016, 19, 1569-1582.	7.1	213
20	Common variants at 12q14 and 12q24 are associated with hippocampal volume. Nature Genetics, 2012, 44, 545-551.	9.4	212
21	Genomeâ€wide association studies of cerebral white matter lesion burden. Annals of Neurology, 2011, 69, 928-939.	2.8	201
22	Genetic architecture of subcortical brain structures in 38,851 individuals. Nature Genetics, 2019, 51, 1624-1636.	9.4	192
23	Whole exome sequencing study identifies novel rare and common Alzheimerâ€™s-Associated variants involved in immune response and transcriptional regulation. Molecular Psychiatry, 2020, 25, 1859-1875.	4.1	191
24	Genetic correlates of brain aging on MRI and cognitive test measures: a genome-wide association and linkage analysis in the Framingham study. BMC Medical Genetics, 2007, 8, S15.	2.1	179
25	Convergent genetic and expression data implicate immunity in Alzheimer's disease. Alzheimer's and Dementia, 2015, 11, 658-671.	0.4	173
26	The Framingham Heart Study 100K SNP genome-wide association study resource: overview of 17 phenotype working group reports. BMC Medical Genetics, 2007, 8, S1.	2.1	169
27	Gene-Wide Analysis Detects Two New Susceptibility Genes for Alzheimer's Disease. PLoS ONE, 2014, 9, e94661.	1.1	155
28	APOE genotype and MRI markers of cerebrovascular disease. Neurology, 2013, 81, 292-300.	1.5	149
29	Influence of Heterozygosity for Parkin Mutation on Onset Age in Familial Parkinson Disease. Archives of Neurology, 2006, 63, 826.	4.9	147
30	Polygenic Overlap Between C-Reactive Protein, Plasma Lipids, and Alzheimer Disease. Circulation, 2015, 131, 2061-2069.	1.6	145
31	Ischemic stroke is associated with the ABO locus: The EuroCLOT study. Annals of Neurology, 2013, 73, 16-31.	2.8	144
32	Common variants in Alzheimerâ€™s disease and risk stratification by polygenic risk scores. Nature Communications, 2021, 12, 3417.	5.8	140
33	Heritability and a Genome-Wide Linkage Scan for Arterial Stiffness, Wave Reflection, and Mean Arterial Pressure. Circulation, 2005, 112, 194-199.	1.6	139
34	Identification of additional risk loci for stroke and small vessel disease: a meta-analysis of genome-wide association studies. Lancet Neurology, The, 2016, 15, 695-707.	4.9	130
35	Common variants at 6q22 and 17q21 are associated with intracranial volume. Nature Genetics, 2012, 44, 539-544.	9.4	126
36	Parental Occurrence of Stroke and Risk of Stroke in Their Children. Circulation, 2010, 121, 1304-1312.	1.6	121

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37	Mutation of FOXC1 and PITX2 induces cerebral small-vessel disease. Journal of Clinical Investigation, 2014, 124, 4877-4881.	3.9	105
38	Genomewide association study for onset age in Parkinson disease. BMC Medical Genetics, 2009, 10, 98.	2.1	104
39	The Gly2019Ser mutation in LRRK2is not fully penetrant in familial Parkinson's disease: the GenePD study. BMC Medicine, 2008, 6, 32.	2.3	102
40	Evidence for a gene influencing the TG/HDL-C ratio on chromosome 7q32.3-qter: a genome-wide scan in the Framingham Study. Human Molecular Genetics, 2000, 9, 1315-1320.	1.4	100
41	PARK3 Influences Age at Onset in Parkinson Disease: A Genome Scan in the GenePD Study. American Journal of Human Genetics, 2002, 70, 1089-1095.	2.6	96
42	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. Neurobiology of Aging, 2015, 36, 1605.e7-1605.e12.	1.5	96
43	Common Variants in the 5â€² Region of the Leptin Gene Are Associated with Body Mass Index in Men from the National Heart, Lung, and Blood Institute Family Heart Study. American Journal of Human Genetics, 2004, 75, 220-230.	2.6	86
44	Genome-Wide Association Studies of MRI-Defined Brain Infarcts. Stroke, 2010, 41, 210-217.	1.0	82
45	Evidence for Linkage Between Essential Hypertension and a Putative Locus on Human Chromosome 17. Hypertension, 1999, 34, 4-7.	1.3	81
46	Genomewide Linkage Analysis to Presbycusis in the Framingham Heart Study. JAMA Otolaryngology, 2003, 129, 285.	1.5	81
47	Evaluation of a Genetic Risk Score to Improve Risk Prediction for Alzheimerâ€™s Disease. Journal of Alzheimer's Disease, 2016, 53, 921-932.	1.2	77
48	Association of NEDD4L Ubiquitin Ligase With Essential Hypertension. Hypertension, 2005, 46, 488-491.	1.3	72
49	Identification of cis - and trans -Acting Genetic Variants Explaining Up to Half the Variation in Circulating Vascular Endothelial Growth Factor Levels. Circulation Research, 2011, 109, 554-563.	2.0	72
50	Correlation between Waardenburg syndrome phenotype and genotype in a population of individuals with identified PAX3 mutations. Human Genetics, 1998, 102, 499-506.	1.8	69
51	Genome-Wide Scan for White Matter Hyperintensity. Stroke, 2006, 37, 77-81.	1.0	67
52	Copy Number Variation in Familial Parkinson Disease. PLoS ONE, 2011, 6, e20988.	1.1	67
53	Genome-wide Studies of Verbal Declarative Memory in Nondemented Older People: The Cohorts for Heart and Aging Research in Genomic Epidemiology Consortium. Biological Psychiatry, 2015, 77, 749-763.	0.7	67
54	Predicting Stroke Through Genetic Risk Functions. Stroke, 2014, 45, 403-412.	1.0	62

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55	Shared genetic contribution to ischemic stroke and Alzheimer's disease. Annals of Neurology, 2016, 79, 739-747.	2.8	56
56	Six Novel Loci Associated with Circulating VEGF Levels Identified by a Meta-analysis of Genome-Wide Association Studies. PLoS Genetics, 2016, 12, e1005874.	1.5	56
57	Genome-Wide Scan for Pulse Pressure in the National Heart, Lung and Blood Instituteâ€™s Framingham Heart Study. Hypertension, 2004, 44, 152-155.	1.3	51
58	Rare and Coding Region Genetic Variants Associated With Risk of Ischemic Stroke. JAMA Neurology, 2015, 72, 781.	4.5	49
59	PLD3 variants in population studies. Nature, 2015, 520, E2-E3.	13.7	49
60	Rare Functional Variant in TM2D3 is Associated with Late-Onset Alzheimer's Disease. PLoS Genetics, 2016, 12, e1006327.	1.5	47
61	Familial paragangliomas: Linkage to chromosome 11q23 and clinical implications. , 1997, 72, 66-70.		46
62	Polymorphisms in the Promoter Region of Catalase Gene and Essential Hypertension. Disease Markers, 2005, 21, 3-7.	0.6	46
63	Autosomal Dominant Orthostatic Hypotensive Disorder Maps to Chromosome 18q. American Journal of Human Genetics, 1998, 63, 1425-1430.	2.6	45
64	A Genome-Wide Scan of Pulmonary Function Measures in the National Heart, Lung, and Blood Institute Family Heart Study. American Journal of Respiratory and Critical Care Medicine, 2003, 167, 1528-1533.	2.5	43
65	Genome-Wide Association Study for Circulating Tissue Plasminogen Activator Levels and Functional Follow-Up Implicates Endothelial STXBP5 and STX2. Arteriosclerosis, Thrombosis, and Vascular Biology, 2014, 34, 1093-1101.	1.1	43
66	Whole genome sequencing of Caribbean Hispanic families with lateâ€onset Alzheimer's disease. Annals of Clinical and Translational Neurology, 2018, 5, 406-417.	1.7	42
67	Maternal component in the familial aggregation of hypertension. Clinical Genetics, 2001, 60, 13-21.	1.0	40
68	Cardiovascular health, genetic risk, and risk of dementia in the Framingham Heart Study. Neurology, 2020, 95, e1341-e1350.	1.5	37
69	Identification of a polymorphic glutamic acid stretch in the Î±2b-adrenergic receptor and lack of linkage with essential hypertension. American Journal of Hypertension, 1999, 12, 853-857.	1.0	36
70	Novel microRNA discovery using small RNA sequencing in post-mortem human brain. BMC Genomics, 2016, 17, 776.	1.2	36
71	Association of polymorphisms in the promoter region of the PNMT gene with essential hypertension in African Americans but not in whites. American Journal of Hypertension, 2003, 16, 859-863.	1.0	34
72	Linkage and association with pulmonary function measures on chromosome 6q27 in the Framingham Heart Study. Human Molecular Genetics, 2003, 12, 2745-2751.	1.4	34

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73	Sequence variation of bradykinin receptors B1 and B2 and association with hypertension. Journal of Hypertension, 2005, 23, 55-62.	0.3	34
74	Replication of association between ELAVL4 and Parkinson disease: the GenePD study. Human Genetics, 2008, 124, 95-99.	1.8	34
75	Whole genome sequence analyses of brain imaging measures in the Framingham Study. Neurology, 2018, 90, e188-e196.	1.5	34
76	Genome-Wide Association Study of Determinants of Anti-Cyclic Citrullinated Peptide Antibody Titer in Adults with Rheumatoid Arthritis. Molecular Medicine, 2009, 15, 136-143.	1.9	33
77	Risk of Parkinson's disease after tamoxifen treatment. BMC Neurology, 2010, 10, 23.	0.8	33
78	Analysis of Whole-Exome Sequencing Data for Alzheimer Disease Stratified by APOE Genotype. JAMA Neurology, 2019, 76, 1099.	4.5	32
79	Genetic Variants of WNK4 in Whites and African Americans With Hypertension. Hypertension, 2003, 41, 1191-1195.	1.3	30
80	Sepiapterin reductase expression is increased in Parkinson's disease brain tissue. Brain Research, 2007, 1139, 42-47.	1.1	30
81	Genetic and lifestyle risk factors for MRI-defined brain infarcts in a population-based setting. Neurology, 2019, 92, .	1.5	30
82	Genetic Predisposition to Stroke in Relatives of Hypertensives. Stroke, 2000, 31, 487-492.	1.0	28
83	Data mining, neural nets, trees â€" Problems 2 and 3 of Genetic Analysis Workshop 15. Genetic Epidemiology, 2007, 31, S51-S60.	0.6	28
84	Pathway analysis following association study. BMC Proceedings, 2011, 5, S18.	1.8	28
85	Is DFNA5 a susceptibility gene for age-related hearing impairment?. European Journal of Human Genetics, 2002, 10, 883-886.	1.4	27
86	Two-stage approach for identifying single-nucleotide polymorphisms associated with rheumatoid arthritis using random forests and Bayesian networks. BMC Proceedings, 2007, 1, S56.	1.8	27
87	Bivariate Heritability of Total and Regional Brain Volumes. Alzheimer Disease and Associated Disorders, 2009, 23, 218-223.	0.6	27
88	Quality control and integration of genotypes from two calling pipelines for whole genome sequence data in the Alzheimer's disease sequencing project. Genomics, 2019, 111, 808-818.	1.3	26
89	Linkage disequilibrium analysis in Machado-Joseph disease patients of different ethnic origins. Human Genetics, 1996, 98, 620-624.	1.8	24
90	Genomeâ€wide linkage analyses of nonâ€Hispanic white families identify novel loci for familial lateâ€onset Alzheimer's disease. Alzheimer's and Dementia, 2016, 12, 2-10.	0.4	24

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91	Genetic Variation in Genes Underlying Diverse Dementias May Explain a Small Proportion of Cases in the Alzheimerâ€™s Disease Sequencing Project. Dementia and Geriatric Cognitive Disorders, 2018, 45, 1-17.	0.7	22
92	Association of HSP70 and its Co-Chaperones with Alzheimer's Disease. Journal of Alzheimer's Disease, 2011, 25, 93-102.	1.2	21
93	Plasma amyloid Î² levels are driven by genetic variants near APOE, BACE1, APP, PSEN2: A genomeâ€wide association study in over 12,000 nonâ€demented participants. Alzheimer's and Dementia, 2021, 17, 1663-1674.	0.4	20
94	A locus for autosomal recessive achromatopsia on human chromosome 8q. Clinical Genetics, 1999, 56, 82-85.	1.0	18
95	Strategies to Design and Analyze Targeted Sequencing Data. Circulation: Cardiovascular Genetics, 2014, 7, 335-343.	5.1	18
96	Exome Chip Analysis Identifies Low-Frequency and Rare Variants in MRPL38 for White Matter Hyperintensities on Brain Magnetic Resonance Imaging. Stroke, 2018, 49, 1812-1819.	1.0	17
97	A novel mutation in the MITF gene causes Waardenburg Syndrome Type 2. Genetic Analysis, Techniques and Applications, 1996, 13, 43-44.	1.5	15
98	Expectation Maximization Algorithm Based Haplotype Relative Risk (EM-HRR): Test of Linkage Disequilibrium Using Incomplete Case-Parents Trios. Human Heredity, 2005, 59, 125-135.	0.4	15
99	Multiomics integrative analysis identifies APOE allele-specific blood biomarkers associated to Alzheimerâ€™s disease etiopathogenesis. Aging, 2021, 13, 9277-9329.	1.4	15
100	HaploBuild: an algorithm to construct non-contiguous associated haplotypes in family based genetic studies. Bioinformatics, 2007, 23, 2190-2192.	1.8	13
101	Informative-Transmission Disequilibrium Test (i-TDT): combined linkage and association mapping that includes unaffected offspring as well as affected offspring. Genetic Epidemiology, 2007, 31, 115-133.	0.6	13
102	Gender equality in Machadoâ€“Joseph disease. Nature Genetics, 1995, 11, 118-119.	9.4	12
103	Combined haplotype relative risk (CHRR): a general and simple genetic association test that combines trios and unrelated caseâ€controls. Genetic Epidemiology, 2009, 33, 54-62.	0.6	12
104	Postmortem Interval InfluencesÎ±-Synuclein Expression in Parkinson Disease Brain. Parkinson's Disease, 2012, 2012, 1-8.	0.6	11
105	Associations of NINJ2 Sequence Variants with Incident Ischemic Stroke in the Cohorts for Heart and Aging in Genomic Epidemiology (CHARGE) Consortium. PLoS ONE, 2014, 9, e99798.	1.1	11
106	Genetic analyses of longitudinal phenotype data: a comparison of univariate methods and a multivariate approach. BMC Genetics, 2003, 4, S29.	2.7	9
107	Huntington CAG repeat size does not modify onset age in familial Parkinson's disease: The GenePD study. Movement Disorders, 2008, 23, 1596-1601.	2.2	8
108	Genomewide linkage study of modifiers of LRRK2â€related Parkinson's disease. Movement Disorders, 2011, 26, 2039-2044.	2.2	8

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109	Estrogen-related and other disease diagnoses preceding Parkinson’s disease. Clinical Epidemiology, 2010, 2, 153.	1.5	7
110	Evaluation of power of the Illumina HumanOmni5M-4v1 BeadChip to detect risk variants for human complex diseases. European Journal of Human Genetics, 2016, 24, 1029-1034.	1.4	7
111	Identifying rare variants from exome scans: the GAW17 experience. BMC Proceedings, 2011, 5, S1.	1.8	6
112	Meta-analysis of genome-wide association studies identifies ancestry-specific associations underlying circulating total tau levels. Communications Biology, 2022, 5, 336.	2.0	6
113	Genetic Interaction with Plasma Lipids on Alzheimerâ€™s Disease in the Framingham Heart Study. Journal of Alzheimer's Disease, 2018, 66, 1275-1282.	1.2	5
114	Integrative methylation score to identify epigenetic modifications associated with lipid changes resulting from fenofibrate treatment in families. BMC Proceedings, 2018, 12, 28.	1.8	5
115	Incorporating biological information into association studies of sequencing data. Genetic Epidemiology, 2011, 35, S29-34.	0.6	4
116	Alzheimer's disease GWAS weighted by multiâ€omics and endophenotypes identifies novel risk loci. Alzheimer's and Dementia, 2020, 16, e043977.	0.4	4
117	Exploiting family history in aggregation unit-based genetic association tests. European Journal of Human Genetics, 2022, 30, 1355-1362.	1.4	4
118	Evaluation of population stratification adjustment using genomeâ€wide or exonic variants. Genetic Epidemiology, 2020, 44, 702-716.	0.6	3
119	Genetically elevated highâ€density lipoprotein cholesterol through the cholesteryl ester transfer protein gene does not associate with risk of Alzheimer's disease. Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring, 2018, 10, 595-598.	1.2	2
120	Detecting linkage for a complex disease using simulated extended pedigrees. Genetic Epidemiology, 1997, 14, 981-986.	0.6	1
121	Power of concordant versus discordant sib pairs at different penetrance levels. Genetic Epidemiology, 1999, 17, S679-84.	0.6	1
122	P1â€018: Rare Deleterious And Lossâ€ofâ€Function Variants in OPRL1 and GAS2L2 Contribute to the Risk of Lateâ€Onset Alzheimerâ€™s Disease: Alzheimerâ€™s Disease Sequencing Project Caseâ€Control Study. Alzheimer's and Dementia, 2016, 12, P406.	0.4	1
123	Genomeâ€wide metaâ€analysis of lateâ€onset Alzheimerâ€™s disease using rare variant imputation in 65,602 subjects identifies risk loci with roles in memory, neurodevelopment, and cardiometabolic traits: The international genomics of Alzheimerâ€™s project (IGAP). Alzheimer's and Dementia, 2020, 16, e044193.	0.4	1
124	Family history aggregation unit-based tests to detect rare genetic variant associations with application to the Framingham Heart Study. American Journal of Human Genetics, 2022, 109, 738-749.	2.6	1
125	Building the Biostatistics Pipeline: Summer Institutes for Training in Biostatistics (SIBS). Chance, 2013, 26, 4-9.	0.1	0
126	F4-04-03: DO THE VARIANTS IDENTIFIED IN IGAP IMPROVE RISK PREDICTION OF ALZHEIMER'S DISEASE?. , 2014, 10, P245-P246.		0

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127	P2â€097: The Alzheimerâ€™s Disease Sequencing Project (ADSP): Data Production, Management, and Availability. Alzheimer's and Dementia, 2016, 12, P648.	0.4	0
128	F1-01-03: Rare Deleterious and Loss-of-Function Variants in OPRL1 and GAS2L2 Contribute to the Risk of Late-Onset Alzheimerâ€™s Disease: Alzheimerâ€™s Disease Sequencing Project Case-Control Study. , 2016, 12, P163-P163.		0
129	O1â€09â€04: Identification of Whole Exome Sequencing Variants Associated with Lateâ€Onset Alzheimer's Disease in the Cohorts for Heart and Aging Research in Genomic Epidemiology (Charge) Consortium. Alzheimer's and Dementia, 2016, 12, P197.	0.4	0
130	S3â€01â€01: Gene Expression, Pathology and Genetic Epidemiology in Large Populationâ€Based Studies. Alzheimer's and Dementia, 2016, 12, P267.	0.4	0
131	[O1â€“11â€“04]: TOPMED WHOLE GENOME SEQUENCE (WGS) ASSOCIATIONS WITH BRAIN MRI MEASURES IN THE FRAMINGHAM STUDY. Alzheimer's and Dementia, 2017, 13, P219.	0.4	0
132	[P3â€“090]: THE ALZHEIMER's DISEASE SEQUENCING PROJECT (ADSP) DATA UPDATE 2017. Alzheimer's and Dementia, 2017, 13, P968.	0.4	0
133	Genetic analysis of biobank data: Familial history aggregationâ€based tests (FHAT) with application to Alzheimer's disease. Alzheimer's and Dementia, 2020, 16, e038648.	0.4	0
134	Whole genome sequence association analyses of brain volumes in the TOPMed program. Alzheimer's and Dementia, 2020, 16, e040627.	0.4	0
135	Comparative transâ€ethnic metaâ€analysis of whole exome sequencing variation for Alzheimerâ€™s disease (AD) in 18,402 individuals of the Alzheimerâ€™s Disease Sequencing Project (ADSP). Alzheimer's and Dementia, 2020, 16, e041583.	0.4	0
136	Assessing whole genome sequencing variation for Alzheimerâ€™s disease in 4707 individuals from the Alzheimerâ€™s Disease Sequencing Project (ADSP). Alzheimer's and Dementia, 2020, 16, e045548.	0.4	0
137	Frequency of familial Alzheimerâ€™s disease gene mutations within the Alzheimer Disease Sequencing Project (ADSP). Alzheimer's and Dementia, 2020, 16, e046203.	0.4	0