Anita L Destefano

List of Publications by Citations

139 papers	16,208 citations	51 h-index	127 g-index
152 ext. papers	19,943 ext. citations	10 avg, IF	4.68 L-index

#	Paper	IF	Citations
139	Meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for Alzheimer's disease. *Nature Genetics*, **2013**, 45, 1452-8	36.3	2714
138	Common variants at ABCA7, MS4A6A/MS4A4E, EPHA1, CD33 and CD2AP are associated with Alzheimer's disease. *Nature Genetics*, **2011**, 43, 429-35	36.3	1421
137	Large-scale meta-analysis of genome-wide association data identifies six new risk loci for Parkinson's disease. *Nature Genetics*, **2014**, 46, 989-93	36.3	1261
136	Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates A tau, immunity and lipid processing. *Nature Genetics*, **2019**, 51, 414-430	36.3	917
135	Genome-wide analysis of genetic loci associated with Alzheimer disease. *JAMA - Journal of the American Medical Association*, **2010**, 303, 1832-40	27.4	888
134	Multiancestry genome-wide association study of 520,000 subjects identifies 32 loci associated with stroke and stroke subtypes. *Nature Genetics*, **2018**, 50, 524-537	36.3	536
133	Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. *Nature Genetics*, **2017**, 49, 1373-1384	36.3	508
132	Evidence for a gene influencing blood pressure on chromosome 17. Genome scan linkage results for longitudinal blood pressure phenotypes in subjects from the framingham heart study. *Hypertension*, **2000**, 36, 477-83	8.5	483
131	Comprehensive research synopsis and systematic meta-analyses in Parkinson's disease genetics: The PDGene database. *PLoS Genetics*, **2012**, 8, e1002548	6	420
130	Genomewide association studies of stroke. *New England Journal of Medicine*, **2009**, 360, 1718-28	59.2	376
129	Genomewide association study for susceptibility genes contributing to familial Parkinson disease. *Human Genetics*, **2009**, 124, 593-605	6.3	363
128	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, 951-62	24.1	359
127	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. *Nature Genetics*, **2011**, 43, 1005-11	36.3	338
126	Shared genetic susceptibility to ischemic stroke and coronary artery disease: a genome-wide analysis of common variants. *Stroke*, **2014**, 45, 24-36	6.7	245
125	A common haplotype lowers PU.1 expression in myeloid cells and delays onset of Alzheimer's disease. *Nature Neuroscience*, **2017**, 20, 1052-1061	25.5	228
124	Meta-analysis of Parkinson's disease: identification of a novel locus, RIT2. *Annals of Neurology*, **2012** , 71, 370-84	9.4	214
123	Common variants at 12q14 and 12q24 are associated with hippocampal volume. *Nature Genetics*, **2012**, 44, 545-51	36.3	175

122	Novel genetic loci associated with hippocampal volume. *Nature Communications*, **2017**, 8, 13624	17.4	173
121	Serum brain-derived neurotrophic factor and the risk for dementia: the Framingham Heart Study. *JAMA Neurology*, **2014**, 71, 55-61	17.2	162
120	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 Suppl 1, S15	2.1	156
119	The Framingham Heart Study 100K SNP genome-wide association study resource: overview of 17 phenotype working group reports. *BMC Medical Genetics*, **2007**, 8 Suppl 1, S1	2.1	152
118	Novel genetic loci underlying human intracranial volume identified through genome-wide association. *Nature Neuroscience*, **2016**, 19, 1569-1582	25.5	147
117	Convergent genetic and expression data implicate immunity in Alzheimer's disease. *Alzheimer's and Dementia*, **2015**, 11, 658-71	1.2	146
116	Genome-wide association studies of cerebral white matter lesion burden: the CHARGE consortium. *Annals of Neurology*, **2011**, 69, 928-39	9.4	146
115	Influence of heterozygosity for parkin mutation on onset age in familial Parkinson disease: the GenePD study. *Archives of Neurology*, **2006**, 63, 826-32		131
114	Heritability and a genome-wide linkage scan for arterial stiffness, wave reflection, and mean arterial pressure: the Framingham Heart Study. *Circulation*, **2005**, 112, 194-9	16.7	121
113	Whole exome sequencing study identifies novel rare and common Alzheimer's-Associated variants involved in immune response and transcriptional regulation. *Molecular Psychiatry*, **2020**, 25, 1859-1875	15.1	106
112	Ischemic stroke is associated with the ABO locus: the EuroCLOT study. *Annals of Neurology*, **2013**, 73, 16-31	9.4	105
111	APOE genotype and MRI markers of cerebrovascular disease: systematic review and meta-analysis. *Neurology*, **2013**, 81, 292-300	6.5	104
110	Common variants at 6q22 and 17q21 are associated with intracranial volume. *Nature Genetics*, **2012** , 44, 539-44	36.3	104
109	Polygenic Overlap Between C-Reactive Protein, Plasma Lipids, and Alzheimer Disease. *Circulation*, **2015**, 131, 2061-2069	16.7	100
108	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	24.1	100
107	Parental occurrence of stroke and risk of stroke in their children: the Framingham study. *Circulation* , **2010**, 121, 1304-12	16.7	97
106	Gene-wide analysis detects two new susceptibility genes for Alzheimer's disease. *PLoS ONE*, **2014**, 9, e94661	3.7	90
105	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, 1315-20	5.6	88

104	PARK3 influences age at onset in Parkinson disease: a genome scan in the GenePD study. *American Journal of Human Genetics*, **2002**, 70, 1089-95	11	81
103	Genetic architecture of subcortical brain structures in 38,851 individuals. *Nature Genetics*, **2019**, 51, 1624-1636	30.5	81
102	Genomewide association study for onset age in Parkinson disease. *BMC Medical Genetics*, **2009**, 10, 98	2.1	78
101	Evidence for linkage between essential hypertension and a putative locus on human chromosome 17. *Hypertension*, **1999**, 34, 4-7	8.5	77
100	Genome-wide association studies of MRI-defined brain infarcts: meta-analysis from the CHARGE Consortium. *Stroke*, **2010**, 41, 210-7	6.7	74
99	Common variants in the 5' 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, 220-30	11	73
98	The Gly2019Ser mutation in LRRK2 is not fully penetrant in familial Parkinson's disease: the GenePD study. *BMC Medicine*, **2008**, 6, 32	11.4	72
97	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. *Neurobiology of Aging*, **2015**, 36, 1605.e7-12	5.6	70
96	Genomewide linkage analysis to presbycusis in the Framingham Heart Study. *JAMA Otolaryngology*, **2003**, 129, 285-9		69
95	Mutation of FOXC1 and PITX2 induces cerebral small-vessel disease. *Journal of Clinical Investigation* , **2014**, 124, 4877-81	15.9	64
94	Genome-wide scan for white matter hyperintensity: the Framingham Heart Study. *Stroke*, **2006**, 37, 77-81	6.7	61
93	Correlation between Waardenburg syndrome phenotype and genotype in a population of individuals with identified PAX3 mutations. *Human Genetics*, **1998**, 102, 499-506	6.3	60
92	Association of NEDD4L ubiquitin ligase with essential hypertension. *Hypertension*, **2005**, 46, 488-91	8.5	60
91	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, 554-63	15.7	57
90	Evaluation of a Genetic Risk Score to Improve Risk Prediction for Alzheimer's Disease. *Journal of Alzheimer's Disease*, **2016**, 53, 921-32	4.3	54
89	Copy number variation in familial Parkinson disease. *PLoS ONE*, **2011**, 6, e20988	3.7	53
88	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, 749-63	7.9	48
87	PLD3 variants in population studies. *Nature*, **2015**, 520, E2-3	50.4	47

86	Predicting stroke through genetic risk functions: the CHARGE Risk Score Project. *Stroke*, **2014**, 45, 403-12	8.7	46
85	Genome-wide scan for pulse pressure in the National Heart, Lung and Blood Institute's Framingham Heart Study. *Hypertension*, **2004**, 44, 152-5	8.5	46
84	Six Novel Loci Associated with Circulating VEGF Levels Identified by a Meta-analysis of Genome-Wide Association Studies. *PLoS Genetics*, **2016**, 12, e1005874	6	43
83	Shared genetic contribution to Ischaemic Stroke and Alzheimer's Disease. *Annals of Neurology*, **2016**, 79, 739-747	9.4	42
82	Autosomal dominant orthostatic hypotensive disorder maps to chromosome 18q. *American Journal of Human Genetics*, **1998**, 63, 1425-30	11	41
81	Familial paragangliomas: linkage to chromosome 11q23 and clinical implications. *American Journal of Medical Genetics Part A*, **1997**, 72, 66-70		40
80	Polymorphisms in the promoter region of catalase gene and essential hypertension. *Disease Markers*, **2005**, 21, 3-7	3.2	40
79	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, 1528-33	19.2	38
78	Rare Functional Variant in TM2D3 is Associated with Late-Onset Alzheimer's Disease. *PLoS Genetics*, **2016**, 12, e1006327	6	38
77	Rare and Coding Region Genetic Variants Associated With Risk of Ischemic Stroke: The NHLBI Exome Sequence Project. *JAMA Neurology*, **2015**, 72, 781-8	17.2	37
76	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, 1093-101	9.4	33
75	Maternal component in the familial aggregation of hypertension. *Clinical Genetics*, **2001**, 60, 13-21	4	33
74	Identification of a polymorphic glutamic acid stretch in the alpha2B-adrenergic receptor and lack of linkage with essential hypertension. *American Journal of Hypertension*, **1999**, 12, 853-7	2.3	32
73	Sequence variation of bradykinin receptors B1 and B2 and association with hypertension. *Journal of Hypertension*, **2005**, 23, 55-62	1.9	31
72	Linkage and association with pulmonary function measures on chromosome 6q27 in the Framingham Heart Study. *Human Molecular Genetics*, **2003**, 12, 2745-51	5.6	29
71	Replication of association between ELAVL4 and Parkinson disease: the GenePD study. *Human Genetics*, **2008**, 124, 95-9	6.3	28
70	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, 859-63	2.3	28
69	Genetic variants of WNK4 in whites and African Americans with hypertension. *Hypertension*, **2003**, 41, 1191-5	8.5	27

68	Genome-wide association study of determinants of anti-cyclic citrullinated peptide antibody titer in adults with rheumatoid arthritis. *Molecular Medicine*, **2009**, 15, 136-43	6.2	26
67	Genetic predisposition to stroke in relatives of hypertensives. *Stroke*, **2000**, 31, 487-92	6.7	26
66	Whole genome sequencing of Caribbean Hispanic families with late-onset Alzheimer's disease. *Annals of Clinical and Translational Neurology*, **2018**, 5, 406-417	5.3	25
65	Risk of Parkinson's disease after tamoxifen treatment. *BMC Neurology*, **2010**, 10, 23	3.1	25
64	Is DFNA5 a susceptibility gene for age-related hearing impairment?. *European Journal of Human Genetics*, **2002**, 10, 883-6	5.3	24
63	Two-stage approach for identifying single-nucleotide polymorphisms associated with rheumatoid arthritis using random forests and Bayesian networks. *BMC Proceedings*, **2007**, 1 Suppl 1, S56	2.3	23
62	Linkage disequilibrium analysis in Machado-Joseph disease patients of different ethnic origins. *Human Genetics*, **1996**, 98, 620-4	6.3	23
61	Common variants in Alzheimer's disease and risk stratification by polygenic risk scores. *Nature Communications*, **2021**, 12, 3417	17.4	23
60	Bivariate heritability of total and regional brain volumes: the Framingham Study. *Alzheimer Disease and Associated Disorders*, **2009**, 23, 218-23	2.5	22
59	Sepiapterin reductase expression is increased in Parkinson's disease brain tissue. *Brain Research*, **2007**, 1139, 42-7	3.7	22
58	Data mining, neural nets, trees--problems 2 and 3 of Genetic Analysis Workshop 15. *Genetic Epidemiology*, **2007**, 31 Suppl 1, S51-60	2.6	21
57	Novel microRNA discovery using small RNA sequencing in post-mortem human brain. *BMC Genomics*, **2016**, 17, 776	4.5	20
56	Whole genome sequence analyses of brain imaging measures in the Framingham Study. *Neurology*, **2018**, 90, e188-e196	6.5	19
55	Analysis of Whole-Exome Sequencing Data for Alzheimer Disease Stratified by APOE Genotype. *JAMA Neurology*, **2019**, 76, 1099-1108	17.2	18
54	Association of HSP70 and its co-chaperones with Alzheimer's disease. *Journal of Alzheimer's Disease* , **2011**, 25, 93-102	4.3	18
53	Genome-wide linkage analyses of non-Hispanic white families identify novel loci for familial late-onset Alzheimer's disease. *Alzheimer's and Dementia*, **2016**, 12, 2-10	1.2	18
52	Genetic and lifestyle risk factors for MRI-defined brain infarcts in a population-based setting. *Neurology*, **2019**,	6.5	17
51	Genetic Variation in Genes Underlying Diverse Dementias May Explain a Small Proportion of Cases in the Alzheimer's Disease Sequencing Project. *Dementia and Geriatric Cognitive Disorders*, **2018**, 45, 1-17	2.6	16

50	Pathway analysis following association study. *BMC Proceedings*, **2011**, 5 Suppl 9, S18	2.3	16
49	Strategies to design and analyze targeted sequencing data: cohorts for Heart and Aging Research in Genomic Epidemiology (CHARGE) Consortium Targeted Sequencing Study. *Circulation: Cardiovascular Genetics*, **2014**, 7, 335-43		15
48	Expectation maximization algorithm based haplotype relative risk (EM-HRR): test of linkage disequilibrium using incomplete case-parents trios. *Human Heredity*, **2005**, 59, 125-35	1.1	15
47	Cardiovascular health, genetic risk, and risk of dementia in the Framingham Heart Study. *Neurology*, **2020**, 95, e1341-e1350	6.5	14
46	Combined haplotype relative risk (CHRR): a general and simple genetic association test that combines trios and unrelated case-controls. *Genetic Epidemiology*, **2009**, 33, 54-62	2.6	12
45	Informative-transmission disequilibrium test (i-TDT): combined linkage and association mapping that includes unaffected offspring as well as affected offspring. *Genetic Epidemiology*, **2007**, 31, 115-33	2.6	12
44	HaploBuild: an algorithm to construct non-contiguous associated haplotypes in family based genetic studies. *Bioinformatics*, **2007**, 23, 2190-2	7.2	12
43	Gender equality in Machado-Joseph disease. *Nature Genetics*, **1995**, 11, 118-9	36.3	12
42	Postmortem Interval Influences -Synuclein Expression in Parkinson Disease Brain. *Parkinson's Disease*, **2012**, 2012, 614212	2.6	11
41	A locus for autosomal recessive achromatopsia on human chromosome 8q. *Clinical Genetics*, **1999**, 56, 82-5	4	11
40	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	6.7	10
39	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	4.3	10
38	A novel mutation in the MITF gene causes Waardenburg syndrome type 2. *Genetic Analysis, Techniques and Applications*, **1996**, 13, 43-4		10
37	Genetic analyses of longitudinal phenotype data: a comparison of univariate methods and a multivariate approach. *BMC Genetics*, **2003**, 4 Suppl 1, S29	2.6	8
36	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	3.7	8
35	Genomewide linkage study of modifiers of LRRK2-related Parkinson's disease. *Movement Disorders*, **2011**, 26, 2039-44	7	7
34	Huntington CAG repeat size does not modify onset age in familial Parkinson's disease: the GenePD study. *Movement Disorders*, **2008**, 23, 1596-601	7	7
33	Estrogen-related and other disease diagnoses preceding Parkinson's disease. *Clinical Epidemiology*, **2010**, 2, 153-70	5.9	6

32	Identifying rare variants from exome scans: the GAW17 experience. *BMC Proceedings*, **2011**, 5 Suppl 9, S1	2.3	5
31	Plasma amyloid β levels are driven by genetic variants near APOE, BACE1, APP, PSEN2: A genome-wide association study in over 12,000 non-demented participants. *Alzheimer's and Dementia*, **2021**, 17, 1663-1674	1.2	5
30	Evaluation of power of the Illumina HumanOmni5M-4v1 BeadChip to detect risk variants for human complex diseases. *European Journal of Human Genetics*, **2016**, 24, 1029-34	5.3	4
29	Incorporating biological information into association studies of sequencing data. *Genetic Epidemiology*, **2011**, 35 Suppl 1, S29-34	2.6	4
28	Multiomics integrative analysis identifies allele-specific blood biomarkers associated to Alzheimer's disease etiopathogenesis. *Aging*, **2021**, 13, 9277-9329	5.6	4
27	Integrative methylation score to identify epigenetic modifications associated with lipid changes resulting from fenofibrate treatment in families. *BMC Proceedings*, **2018**, 12, 28	2.3	4
26	Genetic Interaction with Plasma Lipids on Alzheimer's Disease in the Framingham Heart Study. *Journal of Alzheimer's Disease*, **2018**, 66, 1275-1282	4.3	3
25	Alzheimer's disease GWAS weighted by multi-omics and endophenotypes identifies novel risk loci. *Alzheimer's and Dementia*, **2020**, 16, e043977	1.2	2
24	A common haplotype lowers PU.1 expression in myeloid cells and delays onset of Alzheimer's disease		2
23	Genome-Wide Meta-Analysis of Late-Onset Alzheimer's Disease Using Rare Variant Imputation in 65,602 Subjects Identifies Novel Rare Variant Locus NCK2: The International Genomics of Alzheimer's Project (IGAP)		2
22	Evaluation of population stratification adjustment using genome-wide or exonic variants. *Genetic Epidemiology*, **2020**, 44, 702-716	2.6	1
21	Detecting linkage for a complex disease using simulated extended pedigrees. *Genetic Epidemiology*, **1997**, 14, 981-6	2.6	1
20	Power of concordant versus discordant sib pairs at different penetrance levels. *Genetic Epidemiology*, **1999**, 17 Suppl 1, S679-84	2.6	1
19	Exploiting family history in aggregation unit-based genetic association tests. *European Journal of Human Genetics*, **2021**,	5.3	1
18	Exome sequencing identifies rare damaging variants in the ATP8B4 and ABCA1 genes as novel risk factors for Alzheimer's Disease		1
17	P1-018: Rare Deleterious And Loss-of-Function Variants in OPRL1 and GAS2L2 Contribute to the Risk of Late-Onset Alzheimer's Disease: Alzheimer's Disease Sequencing Project Case-Control Study **2016**, 12, P406-P406		1
16	Genome-wide meta-analysis of late-onset Alzheimer'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 Alzheimer's project (IGAP). *Alzheimer's and Dementia*, **2020**, 16, e044193	1.2	0
15	Meta-analysis of genome-wide association studies identifies ancestry-specific associations underlying circulating total tau levels.. *Communications Biology*, **2022**, 5, 336	6.7	0

14	Genetic analysis of biobank data: Familial history aggregation-based tests (FHAT) with application to Alzheimer's disease. *Alzheimer's and Dementia*, **2020**, 16, e038648	1.2
13	Whole genome sequence association analyses of brain volumes in the TOPMed program. *Alzheimer's and Dementia*, **2020**, 16, e040627	1.2
12	Comparative trans-ethnic meta-analysis of whole exome sequencing variation for Alzheimer's disease (AD) in 18,402 individuals of the Alzheimer's Disease Sequencing Project (ADSP). *Alzheimer's and Dementia*, **2020**, 16, e041583	1.2
11	Assessing whole genome sequencing variation for Alzheimer's disease in 4707 individuals from the Alzheimer's Disease Sequencing Project (ADSP). *Alzheimer's and Dementia*, **2020**, 16, e045548	1.2
10	Frequency of familial Alzheimer's disease gene mutations within the Alzheimer Disease Sequencing Project (ADSP). *Alzheimer's and Dementia*, **2020**, 16, e046203	1.2
9	F4-04-03: DO THE VARIANTS IDENTIFIED IN IGAP IMPROVE RISK PREDICTION OF ALZHEIMER'S DISEASE? **2014**, 10, P245-P246	
8	Building the Biostatistics Pipeline: Summer Institutes for Training in Biostatistics (SIBS). *Chance*, **2013**, 26, 4-9	1
7	[O1□11□04]: TOPMED WHOLE GENOME SEQUENCE (WGS) ASSOCIATIONS WITH BRAIN MRI MEASURES IN THE FRAMINGHAM STUDY **2017**, 13, P219-P220	
6	[P3□090]: THE ALZHEIMER's DISEASE SEQUENCING PROJECT (ADSP) DATA UPDATE 2017 **2017**, 13, P968-P968	
5	P2-097: The Alzheimer's Disease Sequencing Project (ADSP): Data Production, Management, and Availability **2016**, 12, P648-P648	
4	F1-01-03: Rare Deleterious and Loss-of-Function Variants in OPRL1 and GAS2L2 Contribute to the Risk of Late-Onset Alzheimer's Disease: Alzheimer's Disease Sequencing Project Case-Control Study **2016**, 12, P163-P163	
3	O1-09-04: Identification of Whole Exome Sequencing Variants Associated with Late-Onset Alzheimer's Disease in the Cohorts for Heart and Aging Research in Genomic Epidemiology (Charge) Consortium **2016**, 12, P197-P198	
2	S3-01-01: Gene Expression, Pathology and Genetic Epidemiology in Large Population-Based Studies **2016**, 12, P267-P267	
1	Genetically elevated high-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	5.2