Anita L Destefano

List of Publications by Year
in descending order

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137 papers	22,336 citations	29994 54 h-index	14702 127 g-index
152 all docs	152 docs citations	152 times ranked	27176 citing authors

#	Article	IF	Citations
1	Exploiting family history in aggregation unit-based genetic association tests. European Journal of Human Genetics, 2022, 30, 1355-1362.	1.4	4
2	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
3	Meta-analysis of genome-wide association studies identifies ancestry-specific associations underlying circulating total tau levels. Communications Biology, 2022, 5, 336.	2.0	6
4	Multiomics integrative analysis identifies APOE allele-specific blood biomarkers associated to Alzheimerâ€™s disease etiopathogenesis. Aging, 2021, 13, 9277-9329.	1.4	15
5	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
6	Common variants in Alzheimerâ€™s disease and risk stratification by polygenic risk scores. Nature Communications, 2021, 12, 3417.	5.8	140
7	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
8	Cardiovascular health, genetic risk, and risk of dementia in the Framingham Heart Study. Neurology, 2020, 95, e1341-e1350.	1.5	37
9	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
10	Whole genome sequence association analyses of brain volumes in the TOPMed program. Alzheimer's and Dementia, 2020, 16, e040627.	0.4	0
11	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
12	Alzheimer's disease GWAS weighted by multiâ€omics and endophenotypes identifies novel risk loci. Alzheimer's and Dementia, 2020, 16, e043977.	0.4	4
13	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
14	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
15	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
16	Evaluation of population stratification adjustment using genomeâ€wide or exonic variants. Genetic Epidemiology, 2020, 44, 702-716.	0.6	3
17	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
18	Analysis of Whole-Exome Sequencing Data for Alzheimer Disease Stratified by APOE Genotype. JAMA Neurology, 2019, 76, 1099.	4.5	32

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19	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
20	Genetic architecture of subcortical brain structures in 38,851 individuals. Nature Genetics, 2019, 51, 1624-1636.	9.4	192
21	Genetic and lifestyle risk factors for MRI-defined brain infarcts in a population-based setting. Neurology, 2019, 92, .	1.5	30
22	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
23	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
24	Whole genome sequence analyses of brain imaging measures in the Framingham Study. Neurology, 2018, 90, e188-e196.	1.5	34
25	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
26	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
27	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
28	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
29	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
30	Novel genetic loci associated with hippocampal volume. Nature Communications, 2017, 8, 13624.	5.8	250
31	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
32	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
33	[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
34	[P3â€“090]: THE ALZHEIMER's DISEASE SEQUENCING PROJECT (ADSP) DATA UPDATE 2017. Alzheimer's and Dementia, 2017, 13, P968.	0.4	0
35	P2â€097: The Alzheimerâ€™s Disease Sequencing Project (ADSP): Data Production, Management, and Availability. Alzheimer's and Dementia, 2016, 12, P648.	0.4	0
36	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

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37	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
38	S3â€01â€01: Gene Expression, Pathology and Genetic Epidemiology in Large Populationâ€Based Studies. Alzheimer's and Dementia, 2016, 12, P267.	0.4	0
39	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
40	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
41	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
42	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
43	Novel genetic loci underlying human intracranial volume identified through genome-wide association. Nature Neuroscience, 2016, 19, 1569-1582.	7.1	213
44	Shared genetic contribution to ischemic stroke and Alzheimer's disease. Annals of Neurology, 2016, 79, 739-747.	2.8	56
45	Novel microRNA discovery using small RNA sequencing in post-mortem human brain. BMC Genomics, 2016, 17, 776.	1.2	36
46	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
47	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
48	Rare Functional Variant in TM2D3 is Associated with Late-Onset Alzheimer's Disease. PLoS Genetics, 2016, 12, e1006327.	1.5	47
49	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. Neurobiology of Aging, 2015, 36, 1605.e7-1605.e12.	1.5	96
50	Rare and Coding Region Genetic Variants Associated With Risk of Ischemic Stroke. JAMA Neurology, 2015, 72, 781.	4.5	49
51	PLD3 variants in population studies. Nature, 2015, 520, E2-E3.	13.7	49
52	Polygenic Overlap Between C-Reactive Protein, Plasma Lipids, and Alzheimer Disease. Circulation, 2015, 131, 2061-2069.	1.6	145
53	Convergent genetic and expression data implicate immunity in Alzheimer's disease. Alzheimer's and Dementia, 2015, 11, 658-671.	0.4	173
54	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

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55	Serum Brain-Derived Neurotrophic Factor and the Risk for Dementia. JAMA Neurology, 2014, 71, 55.	4.5	219
56	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
57	Predicting Stroke Through Genetic Risk Functions. Stroke, 2014, 45, 403-412.	1.0	62
58	Shared Genetic Susceptibility to Ischemic Stroke and Coronary Artery Disease. Stroke, 2014, 45, 24-36.	1.0	302
59	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
60	Strategies to Design and Analyze Targeted Sequencing Data. Circulation: Cardiovascular Genetics, 2014, 7, 335-343.	5.1	18
61	F4-04-03: DO THE VARIANTS IDENTIFIED IN IGAP IMPROVE RISK PREDICTION OF ALZHEIMER'S DISEASE?. , 2014, 10, P245-P246.		0
62	Mutation of FOXC1 and PITX2 induces cerebral small-vessel disease. Journal of Clinical Investigation, 2014, 124, 4877-4881.	3.9	105
63	Gene-Wide Analysis Detects Two New Susceptibility Genes for Alzheimer's Disease. PLoS ONE, 2014, 9, e94661.	1.1	155
64	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
65	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
66	Building the Biostatistics Pipeline: Summer Institutes for Training in Biostatistics (SIBS). Chance, 2013, 26, 4-9.	0.1	0
67	Ischemic stroke is associated with the ABO locus: The EuroCLOT study. Annals of Neurology, 2013, 73, 16-31.	2.8	144
68	APOE genotype and MRI markers of cerebrovascular disease. Neurology, 2013, 81, 292-300.	1.5	149
69	Comprehensive Research Synopsis and Systematic Meta-Analyses in Parkinson's Disease Genetics: The PDGene Database. PLoS Genetics, 2012, 8, e1002548.	1.5	495
70	Common variants at 6q22 and 17q21 are associated with intracranial volume. Nature Genetics, 2012, 44, 539-544.	9.4	126
71	Common variants at 12q14 and 12q24 are associated with hippocampal volume. Nature Genetics, 2012, 44, 545-551.	9.4	212
72	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

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73	Postmortem Interval InfluencesÎ±-Synuclein Expression in Parkinson Disease Brain. Parkinson's Disease, 2012, 2012, 1-8.	0.6	11
74	Metaâ€analysis of Parkinson's Disease: Identification of a novel locus, RIT2. Annals of Neurology, 2012, 71, 370-384.	2.8	264
75	Association of HSP70 and its Co-Chaperones with Alzheimer's Disease. Journal of Alzheimer's Disease, 2011, 25, 93-102.	1.2	21
76	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
77	Identifying rare variants from exome scans: the GAW17 experience. BMC Proceedings, 2011, 5, S1.	1.8	6
78	Pathway analysis following association study. BMC Proceedings, 2011, 5, S18.	1.8	28
79	Genomewide linkage study of modifiers of LRRK2â€related Parkinson's disease. Movement Disorders, 2011, 26, 2039-2044.	2.2	8
80	Genomeâ€wide association studies of cerebral white matter lesion burden. Annals of Neurology, 2011, 69, 928-939.	2.8	201
81	Incorporating biological information into association studies of sequencing data. Genetic Epidemiology, 2011, 35, S29-34.	0.6	4
82	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
83	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. Nature Genetics, 2011, 43, 1005-1011.	9.4	403
84	Copy Number Variation in Familial Parkinson Disease. PLoS ONE, 2011, 6, e20988.	1.1	67
85	Genome-Wide Association Studies of MRI-Defined Brain Infarcts. Stroke, 2010, 41, 210-217.	1.0	82
86	Risk of Parkinson's disease after tamoxifen treatment. BMC Neurology, 2010, 10, 23.	0.8	33
87	Estrogen-related and other disease diagnoses preceding Parkinson’s disease. Clinical Epidemiology, 2010, 2, 153.	1.5	7
88	Genome-wide Analysis of Genetic Loci Associated With Alzheimer Disease. JAMA - Journal of the American Medical Association, 2010, 303, 1832.	3.8	1,064
89	Parental Occurrence of Stroke and Risk of Stroke in Their Children. Circulation, 2010, 121, 1304-1312.	1.6	121
90	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

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91	Genomewide Association Studies of Stroke. New England Journal of Medicine, 2009, 360, 1718-1728.	13.9	420
92	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
93	Genomewide association study for susceptibility genes contributing to familial Parkinson disease. Human Genetics, 2009, 124, 593-605.	1.8	410
94	Genomewide association study for onset age in Parkinson disease. BMC Medical Genetics, 2009, 10, 98.	2.1	104
95	Bivariate Heritability of Total and Regional Brain Volumes. Alzheimer Disease and Associated Disorders, 2009, 23, 218-223.	0.6	27
96	Replication of association between ELAVL4 and Parkinson disease: the GenePD study. Human Genetics, 2008, 124, 95-99.	1.8	34
97	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
98	The Gly2019Ser mutation in LRRK2is not fully penetrant in familial Parkinson's disease: the GenePD study. BMC Medicine, 2008, 6, 32.	2.3	102
99	HaploBuild: an algorithm to construct non-contiguous associated haplotypes in family based genetic studies. Bioinformatics, 2007, 23, 2190-2192.	1.8	13
100	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
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	Data mining, neural nets, trees â€" Problems 2 and 3 of Genetic Analysis Workshop 15. Genetic Epidemiology, 2007, 31, S51-S60.	0.6	28
103	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
104	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
105	Sepiapterin reductase expression is increased in Parkinson's disease brain tissue. Brain Research, 2007, 1139, 42-47.	1.1	30
106	Influence of Heterozygosity for Parkin Mutation on Onset Age in Familial Parkinson Disease. Archives of Neurology, 2006, 63, 826.	4.9	147
107	Genome-Wide Scan for White Matter Hyperintensity. Stroke, 2006, 37, 77-81.	1.0	67
108	Sequence variation of bradykinin receptors B1 and B2 and association with hypertension. Journal of Hypertension, 2005, 23, 55-62.	0.3	34

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109	Polymorphisms in the Promoter Region of Catalase Gene and Essential Hypertension. Disease Markers, 2005, 21, 3-7.	0.6	46
110	Heritability and a Genome-Wide Linkage Scan for Arterial Stiffness, Wave Reflection, and Mean Arterial Pressure. Circulation, 2005, 112, 194-199.	1.6	139
111	Association of NEDD4L Ubiquitin Ligase With Essential Hypertension. Hypertension, 2005, 46, 488-491.	1.3	72
112	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
113	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
114	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
115	Genetic analyses of longitudinal phenotype data: a comparison of univariate methods and a multivariate approach. BMC Genetics, 2003, 4, S29.	2.7	9
116	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
117	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
118	Genetic Variants of WNK4 in Whites and African Americans With Hypertension. Hypertension, 2003, 41, 1191-1195.	1.3	30
119	Genomewide Linkage Analysis to Presbycusis in the Framingham Heart Study. JAMA Otolaryngology, 2003, 129, 285.	1.5	81
120	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
121	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
122	Is DFNA5 a susceptibility gene for age-related hearing impairment?. European Journal of Human Genetics, 2002, 10, 883-886.	1.4	27
123	Maternal component in the familial aggregation of hypertension. Clinical Genetics, 2001, 60, 13-21.	1.0	40
124	Genetic Predisposition to Stroke in Relatives of Hypertensives. Stroke, 2000, 31, 487-492.	1.0	28
125	Evidence for a Gene Influencing Blood Pressure on Chromosome 17. Hypertension, 2000, 36, 477-483.	1.3	534
126	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

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127	Evidence for Linkage Between Essential Hypertension and a Putative Locus on Human Chromosome 17. Hypertension, 1999, 34, 4-7.	1.3	81
128	A locus for autosomal recessive achromatopsia on human chromosome 8q. Clinical Genetics, 1999, 56, 82-85.	1.0	18
129	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
130	Power of concordant versus discordant sib pairs at different penetrance levels. Genetic Epidemiology, 1999, 17, S679-84.	0.6	1
131	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
132	Autosomal Dominant Orthostatic Hypotensive Disorder Maps to Chromosome 18q. American Journal of Human Genetics, 1998, 63, 1425-1430.	2.6	45
133	Familial paragangliomas: Linkage to chromosome 11q23 and clinical implications. , 1997, 72, 66-70.		46
134	Detecting linkage for a complex disease using simulated extended pedigrees. Genetic Epidemiology, 1997, 14, 981-986.	0.6	1
135	A novel mutation in the MITF gene causes Waardenburg Syndrome Type 2. Genetic Analysis, Techniques and Applications, 1996, 13, 43-44.	1.5	15
136	Linkage disequilibrium analysis in Machado-Joseph disease patients of different ethnic origins. Human Genetics, 1996, 98, 620-624.	1.8	24
137	Gender equality in Machadoâ€“Joseph disease. Nature Genetics, 1995, 11, 118-119.	9.4	12