Amalia Bruni

List of Publications by Year
in descending order

91 papers	8,041 citations	136950 32 h-index	54911 84 g-index
93 all docs	93 docs citations	93 times ranked	11814 citing authors

#	Article	IF	Citations
1	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.	21.4	1,962
2	The neuronal sortilin-related receptor SORL1 is genetically associated with Alzheimer disease. Nature Genetics, 2007, 39, 168-177.	21.4	1,045
3	Nicastrin modulates presenilin-mediated notch/glp-1 signal transduction and Î²APP processing. Nature, 2000, 407, 48-54.	27.8	895
4	Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. Nature Genetics, 2017, 49, 1373-1384.	21.4	783
5	Frontotemporal dementia and its subtypes: a genome-wide association study. Lancet Neurology, The, 2014, 13, 686-699.	10.2	302
6	SQSTM1 mutations in frontotemporal lobar degeneration and amyotrophic lateral sclerosis. Neurology, 2012, 79, 1556-1562.	1.1	252
7	Circulating Levels of Soluble Receptor for Advanced Glycation End Products in Alzheimer Disease and Vascular Dementia. Archives of Neurology, 2005, 62, 1734.	4.5	195
8	Mitochondrial DNA haplogroups and APOE4 allele are non-independent variables in sporadic Alzheimer's disease. Human Genetics, 2001, 108, 194-198.	3.8	154
9	Global Prevalence of Young-Onset Dementia. JAMA Neurology, 2021, 78, 1080.	9.0	124
10	An Î±-2-macroglobulin insertion-deletion polymorphism in Alzheimer disease. Nature Genetics, 1999, 22, 19-21.	21.4	115
11	The C9orf72 repeat expansion itself is methylated in ALS and FTLD patients. Acta Neuropathologica, 2015, 129, 715-727.	7.7	114
12	Autosomal Dominant Frontotemporal Lobar Degeneration Due to the C9ORF72 Hexanucleotide Repeat Expansion: Late-Onset Psychotic Clinical Presentation. Biological Psychiatry, 2013, 74, 384-391.	1.3	105
13	Cathepsin F mutations cause Type B Kufs disease, an adult-onset neuronal ceroid lipofuscinosis. Human Molecular Genetics, 2013, 22, 1417-1423.	2.9	105
14	Potential genetic modifiers of disease risk and age at onset in patients with frontotemporal lobar degeneration and GRN mutations: a genome-wide association study. Lancet Neurology, The, 2018, 17, 548-558.	10.2	97
15	Investigation of C9orf72 in 4 Neurodegenerative Disorders. Archives of Neurology, 2012, 69, 1583.	4.5	89
16	Mutation analysis ofCHCHD10in different neurodegenerative diseases. Brain, 2015, 138, e380-e380.	7.6	86
17	Behavioral Disorder, Dementia, Ataxia, and Rigidity in a Large Family With TATA Box-Binding Protein Mutation. Archives of Neurology, 2004, 61, 1314-20.	4.5	82
18	Hypermethylation of the CpG-island near the C9orf72 G4C2-repeat expansion in FTLD patients. Human Molecular Genetics, 2014, 23, 5630-5637.	2.9	74

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19	PML nuclear bodies and neuronal intranuclear inclusion in polyglutamine diseases. Neurobiology of Disease, 2003, 13, 230-237.	4.4	57
20	NGF controls APP cleavage by downregulating APP phosphorylation at Thr668: relevance for Alzheimer's disease. Aging Cell, 2016, 15, 661-672.	6.7	57
21	Chromosome 9p21.3 genotype is associated with vascular dementia and Alzheimer's disease. Neurobiology of Aging, 2011, 32, 1231-1235.	3.1	56
22	Novel PSEN1 and PGRN mutations in early-onset familial frontotemporal dementia. Neurobiology of Aging, 2009, 30, 1825-1833.	3.1	48
23	A genome-wide screening and SNPs-to-genes approach to identify novel genetic risk factors associated with frontotemporal dementia. Neurobiology of Aging, 2015, 36, 2904.e13-2904.e26.	3.1	48
24	The mitochondrial DNA control region shows genetically correlated levels of heteroplasmy in leukocytes of centenarians and their offspring. BMC Genomics, 2007, 8, 293.	2.8	44
25	Comprehensive Evaluation of Plasma 7-Ketocholesterol and Cholestan-3Î²,5Î±,6Î²-Triol in an Italian Cohort of Patients Affected by Niemann-Pick Disease due to NPC1 and SMPD1 Mutations. Clinica Chimica Acta, 2016, 455, 39-45.	1.1	42
26	APOE polymorphism affects episodic memory among non demented elderly subjects. Experimental Gerontology, 2009, 44, 224-227.	2.8	41
27	A New Italian Pedigree with Early-Onset Alzheimer's Disease. Journal of Geriatric Psychiatry and Neurology, 1994, 7, 28-32.	2.3	40
28	Epidemiology and genetics of frontotemporal dementia: a door-to-door survey in Southern Italy. Neurobiology of Aging, 2012, 33, 2948.e1-2948.e10.	3.1	40
29	Haptoglobin Interacts with Apolipoprotein E and Beta-Amyloid and Influences Their Crosstalk. ACS Chemical Neuroscience, 2014, 5, 837-847.	3.5	39
30	A C6orf10/LOC101929163 locus is associated with age of onset in C9orf72 carriers. Brain, 2018, 141, 2895-2907.	7.6	39
31	Clinical Features Associated with Delirium Motor Subtypes in Older Inpatients: Results of a Multicenter Study. American Journal of Geriatric Psychiatry, 2017, 25, 1064-1071.	1.2	38
32	Brainâ€derived neurotrophic factor modulates cholesterol homeostasis and Apolipoprotein E synthesis in human cell models of astrocytes and neurons. Journal of Cellular Physiology, 2018, 233, 6925-6943.	4.1	33
33	From beta amyloid to altered proteostasis in Alzheimerâ€™s disease. Ageing Research Reviews, 2020, 64, 101126.	10.9	31
34	Homozygous carriers of APP A713T mutation in an autosomal dominant Alzheimer disease family. Neurology, 2015, 84, 2266-2273.	1.1	30
35	Tau Mutations Serve as a Novel Risk Factor for Cancer. Cancer Research, 2018, 78, 3731-3739.	0.9	30
36	Somatic Point Mutations in mtDNA Control Region Are Influenced by Genetic Background and Associated with Healthy Aging: A GEHA Study. PLoS ONE, 2010, 5, e13395.	2.5	28

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37	Clinical Manifestations of Highly Prevalent Corticosteroid-Binding Globulin Mutations in a Village in Southern Italy. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E1684-E1693.	3.6	26
38	Contribution of polymorphic variation of inositol hexakisphosphate kinase 3 (IP6K3) gene promoter to the susceptibility to late onset Alzheimer's disease. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2016, 1862, 1766-1773.	3.8	26
39	Presenilin 2 Ser130Leu mutation in a case of late-onset â€œsporadicâ€•Alzheimerâ€™s disease. Journal of Neurology, 2007, 254, 391-393.	3.6	25
40	Role of Niemann-Pick Type C Disease Mutations in Dementia. Journal of Alzheimer's Disease, 2016, 55, 1249-1259.	2.6	24
41	AÎ²PP A713T Mutation in Late Onset Alzheimer's Disease with Cerebrovascular Lesions. Journal of Alzheimer's Disease, 2009, 17, 383-389.	2.6	23
42	Characterization of nigrostriatal dysfunction in spinocerebellar ataxia 17. Movement Disorders, 2006, 21, 872-875.	3.9	22
43	Relation of Apolipoprotein(a) Size to Alzheimerâ€™s Disease and Vascular Dementia. Dementia and Geriatric Cognitive Disorders, 2004, 18, 189-196.	1.5	21
44	Presenilin-1 mutation E318G and familial Alzheimer's disease in the Italian population. Neurobiology of Aging, 2007, 28, 1682-1688.	3.1	21
45	A novel network analysis approach reveals DNA damage, oxidative stress and calcium/cAMP homeostasis-associated biomarkers in frontotemporal dementia. PLoS ONE, 2017, 12, e0185797.	2.5	21
46	The novel PSEN1 M84V mutation associated to frontal dysexecutive syndrome, spastic paraparesis, and cerebellar atrophy in a dominant Alzheimer's disease family. Neurobiology of Aging, 2017, 56, 213.e7-213.e12.	3.1	19
47	Genetic Counseling and Testing for Alzheimerâ€™s Disease and Frontotemporal Lobar Degeneration: An Italian Consensus Protocol. Journal of Alzheimer's Disease, 2016, 51, 277-291.	2.6	18
48	Mutations in Prion Protein Gene: Pathogenic Mechanisms in C-Terminal vs. N-Terminal Domain, a Review. International Journal of Molecular Sciences, 2019, 20, 3606.	4.1	18
49	Late onset familial Alzheimer's disease: novel presenilin 2 mutation and PS1 E318G polymorphism. Journal of Neurology, 2008, 255, 604-606.	3.6	17
50	The Added Value of Neuropsychologic Tests and Structural Imaging for the Etiologic Diagnosis of Dementia in Italian Expert Centers. Alzheimer Disease and Associated Disorders, 2008, 22, 309-320.	1.3	17
51	Somatic comorbidities and Alzheimerâ€™s disease treatment. Neurological Sciences, 2013, 34, 1581-1589.	1.9	17
52	Apolipoprotein(a) null phenotype is related to a delayed age at onset of Alzheimer's disease. Neuroscience Letters, 2004, 357, 45-48.	2.1	15
53	Novel MAPT Val75Ala mutation and PSEN2 Arg62Hys in two siblings with frontotemporal dementia. Neurological Sciences, 2010, 31, 65-70.	1.9	15
54	The first deep intronic mutation in the NOTCH3 gene in a family with late-onset CADASIL. Neurobiology of Aging, 2013, 34, 2234.e9-2234.e12.	3.1	15

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55	Genetics in Degenerative Dementia. Alzheimer Disease and Associated Disorders, 2014, 28, 199-205.	1.3	15
56	Novel N-terminal domain mutation in prion protein detected in 2 patients diagnosed with frontotemporal lobar degeneration syndrome. Neurobiology of Aging, 2014, 35, 2657.e7-2657.e11.	3.1	15
57	The Genetic Variability of UCP4 Affects the Individual Susceptibility to Late-Onset Alzheimerâ€™s Disease and Modifies the Diseaseâ€™s Risk in APOE-É›4 Carriers. Journal of Alzheimer's Disease, 2016, 51, 1265-1274.	2.6	15
58	Nicastrin gene in familial and sporadic Alzheimer's disease. Neuroscience Letters, 2003, 353, 61-65.	2.1	14
59	A Novel Pathogenic PSEN1 Mutation in a Family with Alzheimer's Disease: Phenotypical and Neuropathological Features. Journal of Alzheimer's Disease, 2011, 25, 425-431.	2.6	14
60	Compound heterozygosity of 2 novel MAPT mutations in frontotemporal dementia. Neurobiology of Aging, 2011, 32, 757.e1-757.e11.	3.1	13
61	MAPT V363I Variation in a Sporadic Case of Frontotemporal Dementia. Alzheimer Disease and Associated Disorders, 2011, 25, 96-99.	1.3	13
62	Amyloid Precursor Protein A713T Mutation in Calabrian Patients with Alzheimerâ€™s Disease: A Population Genomics Approach to Estimate Inheritance from a Common Ancestor. Biomedicines, 2022, 10, 20.	3.2	13
63	Association between small apolipoprotein(a) isoforms and frontotemporal dementia in humans. Neuroscience Letters, 2003, 353, 201-204.	2.1	12
64	Association studies between the plasmin genes and late-onset Alzheimer's disease. Neurobiology of Aging, 2007, 28, 1041-1043.	3.1	12
65	Role of TOMM40 rs10524523 Polymorphism in Onset of Alzheimer's Disease Caused by the PSEN1 M146L Mutation. Journal of Alzheimer's Disease, 2013, 37, 285-289.	2.6	12
66	Prevalence of Delirium in a Population of Elderly Outpatients with Dementia: A Retrospective Study. Journal of Alzheimer's Disease, 2017, 61, 251-257.	2.6	12
67	Absence of association between Alzheimer disease and the regulatory region polymorphism of the PS2 gene in an Italian population. Neuroscience Letters, 2003, 343, 210-212.	2.1	11
68	Association of the Variant Cys139Arg at GRN Gene to the Clinical Spectrum of Frontotemporal Lobar Degeneration. Journal of Alzheimer's Disease, 2014, 40, 679-685.	2.6	11
69	Short-Term Response is not Predictive ofÂLong-Term Response toÂAcetylcholinesterase Inhibitors in Old Age Subjects with Alzheimerâ€™s Disease: AÂâ€œReal Worldâ€•Study. Journal of Alzheimer's Disease, 2017, 56, 239-248.	2.6	11
70	Gene Expression Imputation Across Multiple Tissue Types Provides Insight Into the Genetic Architecture of Frontotemporal Dementia and Its Clinical Subtypes. Biological Psychiatry, 2021, 89, 825-835.	1.3	10
71	Uncoupling protein 4 (UCP4) gene variability in neurodegenerative disorders: further evidence of association in Frontotemporal dementia. Aging, 2018, 10, 3283-3293.	3.1	10
72	IP6K3 and IPMK variations in LOAD and longevity: Evidence for a multifaceted signaling network at the crossroad between neurodegeneration and survival. Mechanisms of Ageing and Development, 2021, 195, 111439.	4.6	9

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73	PSEN1 and PRNP Gene Mutations Co-occurrence Makes Onset Very Early in a Family with FTD Phenotype. Journal of Alzheimer's Disease, 2011, 24, 415-419.	2.6	8
74	No evidence for tau duplications in frontal temporal dementia families showing genetic linkage to the tau locus in which tau mutations have not been found. Neuroscience Letters, 2004, 363, 99-101.	2.1	7
75	Influence of controlled encoding and retrieval facilitation on memory performance in patients with different profiles of mild cognitive impairment. Journal of Neurology, 2015, 262, 938-948.	3.6	7
76	C9orf72, age at onset, and ancestry help discriminate behavioral from language variants in FTLD cohorts. Neurology, 2020, 95, e3288-e3302.	1.1	7
77	SLITRK2, an X-linked modifier of the age at onset in C9orf72 frontotemporal lobar degeneration. Brain, 2021, 144, 2798-2811.	7.6	7
78	Position paper of the Italian Society for the study of Dementias (Sindem) on the proposal of a new Lexicon on Alzheimer disease. Neurological Sciences, 2012, 33, 201-208.	1.9	6
79	Alzheimer's Disease: A Model From the Quantitative Study of a Large Kindred. Topics in Geriatrics, 1992, 5, 126-131.	0.8	5
80	Identification of Three Novel LRRK2 Mutations associated with Parkinson's Disease in a Calabrian Population. Journal of Alzheimer's Disease, 2013, 38, 351-357.	2.6	5
81	Frequency of Cardiovascular Genetic Risk Factors in a Calabrian Population and Their Effects on Dementia. Journal of Alzheimer's Disease, 2018, 61, 1179-1187.	2.6	5
82	The largest caucasian kindred with dentatorubralâ€pallidoluysian atrophy: A founder mutation in italy. Movement Disorders, 2019, 34, 1919-1924.	3.9	5
83	Effects of Multiple Genetic Loci on Age atÂOnset in Frontotemporal Dementia. Journal of Alzheimer's Disease, 2017, 56, 1271-1278.	2.6	4
84	Pathogenic Mechanisms of the Prion Protein Gene Mutations: A Review and Speculative Hypotheses for Pathogenic Potential of the Pro39Leu Mutation in the Associated FTD-Like Phenotype. Journal of Neurology and Neuroscience, 2017, 08, .	0.4	4
85	Mendelian randomization implies no direct causal association between leukocyte telomere length and amyotrophic lateral sclerosis. Scientific Reports, 2020, 10, 12184.	3.3	4
86	Prevalence of heterozygous mutations in Niemann-Pick type C genes in a cohort of progressive supranuclear palsy. Parkinsonism and Related Disorders, 2020, 79, 9-10.	2.2	4
87	Manic depressive illness in a founder population. European Journal of Human Genetics, 2003, 11, 597-602.	2.8	3
88	Angela R.: a familial Alzheimerâ€™s disease case in the days of Auguste D.. Journal of Neurology, 2016, 263, 2494-2498.	3.6	3
89	Comparison of Clinical and Pathological Phenotypes in Two Ethnically and Geographically Unrelated Pedigrees Segregating an Equivalent Presenilin 1 Mutation. Journal of Neuropsychiatry and Clinical Neurosciences, 2000, 12, 359-363.	1.8	2
90	Nicastrin gene in familial and sporadic Alzheimer's disease. Neuroscience Letters, 2003, 353, 61-61.	2.1	1

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91	ALPHA: an eAsy inteLligent service Platform for Healthy Ageing. Biosystems and Biorobotics, 2015, , 419-427.	0.3	0