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	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
2	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
3	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
4	Global Prevalence of Young-Onset Dementia. JAMA Neurology, 2021, 78, 1080.	9.0	124
5	SLITRK2, an X-linked modifier of the age at onset in C9orf72 frontotemporal lobar degeneration. Brain, 2021, 144, 2798-2811.	7.6	7
6	From beta amyloid to altered proteostasis in Alzheimerâ€™s disease. Ageing Research Reviews, 2020, 64, 101126.	10.9	31
7	Mendelian randomization implies no direct causal association between leukocyte telomere length and amyotrophic lateral sclerosis. Scientific Reports, 2020, 10, 12184.	3.3	4
8	C9orf72, age at onset, and ancestry help discriminate behavioral from language variants in FTLD cohorts. Neurology, 2020, 95, e3288-e3302.	1.1	7
9	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
10	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
11	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
12	The largest caucasian kindred with dentatorubralâ€pallidoluysian atrophy: A founder mutation in italy. Movement Disorders, 2019, 34, 1919-1924.	3.9	5
13	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
14	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
15	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
16	A C6orf10/LOC101929163 locus is associated with age of onset in C9orf72 carriers. Brain, 2018, 141, 2895-2907.	7.6	39
17	Tau Mutations Serve as a Novel Risk Factor for Cancer. Cancer Research, 2018, 78, 3731-3739.	0.9	30
18	Uncoupling protein 4 (UCP4) gene variability in neurodegenerative disorders: further evidence of association in Frontotemporal dementia. Aging, 2018, 10, 3283-3293.	3.1	10

#	Article	IF	Citations
19	Effects of Multiple Genetic Loci on Age atÂOnset in Frontotemporal Dementia. Journal of Alzheimer's Disease, 2017, 56, 1271-1278.	2.6	4
20	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
21	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
22	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
23	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
24	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
25	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
26	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
27	Role of Niemann-Pick Type C Disease Mutations in Dementia. Journal of Alzheimer's Disease, 2016, 55, 1249-1259.	2.6	24
28	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
29	Angela R.: a familial Alzheimerâ€™s disease case in the days of Auguste D.. Journal of Neurology, 2016, 263, 2494-2498.	3.6	3
30	NGF controls APP cleavage by downregulating APP phosphorylation at Thr668: relevance for Alzheimer's disease. Aging Cell, 2016, 15, 661-672.	6.7	57
31	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
32	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
33	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
34	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
35	The C9orf72 repeat expansion itself is methylated in ALS and FTLD patients. Acta Neuropathologica, 2015, 129, 715-727.	7.7	114
36	Homozygous carriers of APP A713T mutation in an autosomal dominant Alzheimer disease family. Neurology, 2015, 84, 2266-2273.	1.1	30

#	Article	IF	Citations
37	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
38	Mutation analysis ofCHCHD10in different neurodegenerative diseases. Brain, 2015, 138, e380-e380.	7.6	86
39	ALPHA: an eAsy inteLligent service Platform for Healthy Ageing. Biosystems and Biorobotics, 2015, , 419-427.	0.3	0
40	Hypermethylation of the CpG-island near the C9orf72 G4C2-repeat expansion in FTLD patients. Human Molecular Genetics, 2014, 23, 5630-5637.	2.9	74
41	Genetics in Degenerative Dementia. Alzheimer Disease and Associated Disorders, 2014, 28, 199-205.	1.3	15
42	Haptoglobin Interacts with Apolipoprotein E and Beta-Amyloid and Influences Their Crosstalk. ACS Chemical Neuroscience, 2014, 5, 837-847.	3.5	39
43	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
44	Frontotemporal dementia and its subtypes: a genome-wide association study. Lancet Neurology, The, 2014, 13, 686-699.	10.2	302
45	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
46	Somatic comorbidities and Alzheimerâ€™s disease treatment. Neurological Sciences, 2013, 34, 1581-1589.	1.9	17
47	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
48	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
49	Cathepsin F mutations cause Type B Kufs disease, an adult-onset neuronal ceroid lipofuscinosis. Human Molecular Genetics, 2013, 22, 1417-1423.	2.9	105
50	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
51	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
52	SQSTM1 mutations in frontotemporal lobar degeneration and amyotrophic lateral sclerosis. Neurology, 2012, 79, 1556-1562.	1.1	252
53	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
54	Investigation of C9orf72 in 4 Neurodegenerative Disorders. Archives of Neurology, 2012, 69, 1583.	4.5	89

#	Article	IF	Citations
55	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
56	Chromosome 9p21.3 genotype is associated with vascular dementia and Alzheimer's disease. Neurobiology of Aging, 2011, 32, 1231-1235.	3.1	56
57	Compound heterozygosity of 2 novel MAPT mutations in frontotemporal dementia. Neurobiology of Aging, 2011, 32, 757.e1-757.e11.	3.1	13
58	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
59	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
60	MAPT V363I Variation in a Sporadic Case of Frontotemporal Dementia. Alzheimer Disease and Associated Disorders, 2011, 25, 96-99.	1.3	13
61	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
62	Novel MAPT Val75Ala mutation and PSEN2 Arg62Hys in two siblings with frontotemporal dementia. Neurological Sciences, 2010, 31, 65-70.	1.9	15
63	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
64	APOE polymorphism affects episodic memory among non demented elderly subjects. Experimental Gerontology, 2009, 44, 224-227.	2.8	41
65	AÎ²PP A713T Mutation in Late Onset Alzheimer's Disease with Cerebrovascular Lesions. Journal of Alzheimer's Disease, 2009, 17, 383-389.	2.6	23
66	Novel PSEN1 and PGRN mutations in early-onset familial frontotemporal dementia. Neurobiology of Aging, 2009, 30, 1825-1833.	3.1	48
67	Late onset familial Alzheimer's disease: novel presenilin 2 mutation and PS1 E318G polymorphism. Journal of Neurology, 2008, 255, 604-606.	3.6	17
68	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
69	Association studies between the plasmin genes and late-onset Alzheimer's disease. Neurobiology of Aging, 2007, 28, 1041-1043.	3.1	12
70	Presenilin-1 mutation E318G and familial Alzheimer's disease in the Italian population. Neurobiology of Aging, 2007, 28, 1682-1688.	3.1	21
71	The neuronal sortilin-related receptor SORL1 is genetically associated with Alzheimer disease. Nature Genetics, 2007, 39, 168-177.	21.4	1,045
72	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

#	Article	IF	Citations
73	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
74	Characterization of nigrostriatal dysfunction in spinocerebellar ataxia 17. Movement Disorders, 2006, 21, 872-875.	3.9	22
75	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
76	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
77	Relation of Apolipoprotein(a) Size to Alzheimerâ€™s Disease and Vascular Dementia. Dementia and Geriatric Cognitive Disorders, 2004, 18, 189-196.	1.5	21
78	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
79	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
80	Manic depressive illness in a founder population. European Journal of Human Genetics, 2003, 11, 597-602.	2.8	3
81	Nicastrin gene in familial and sporadic Alzheimer's disease. Neuroscience Letters, 2003, 353, 61-65.	2.1	14
82	Association between small apolipoprotein(a) isoforms and frontotemporal dementia in humans. Neuroscience Letters, 2003, 353, 201-204.	2.1	12
83	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
84	PML nuclear bodies and neuronal intranuclear inclusion in polyglutamine diseases. Neurobiology of Disease, 2003, 13, 230-237.	4.4	57
85	Nicastrin gene in familial and sporadic Alzheimer's disease. Neuroscience Letters, 2003, 353, 61-61.	2.1	1
86	Mitochondrial DNA haplogroups and APOE4 allele are non-independent variables in sporadic Alzheimer's disease. Human Genetics, 2001, 108, 194-198.	3.8	154
87	Nicastrin modulates presenilin-mediated notch/glp-1 signal transduction and Î²APP processing. Nature, 2000, 407, 48-54.	27.8	895
88	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
89	An Î±-2-macroglobulin insertion-deletion polymorphism in Alzheimer disease. Nature Genetics, 1999, 22, 19-21.	21.4	115
90	A New Italian Pedigree with Early-Onset Alzheimer's Disease. Journal of Geriatric Psychiatry and Neurology, 1994, 7, 28-32.	2.3	40

#	Article	IF	Citations
91	Alzheimer's Disease: A Model From the Quantitative Study of a Large Kindred. Topics in Geriatrics, 1992, 5, 126-131.	0.8	5