Stefania Battistini

List of Publications by Year in descending order

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Version: 2024-02-01

61 papers	8,681 citations	218677 26 h-index	149698 56 g-index
62 all docs	62 docs citations	62 times ranked	9350 citing authors

#	Article	IF	Citations
1	BDNF and Pro-BDNF in Amyotrophic Lateral Sclerosis: A New Perspective for Biomarkers of Neurodegeneration. Brain Sciences, 2022, 12, 617.	2.3	8
2	Molecular Genetic Features of Cerebral Cavernous Malformations (CCM) Patients: An Overall View from Genes to Endothelial Cells. Cells, 2021, 10, 704.	4.1	21
3	KRIT1 Gene in Patients with Cerebral Cavernous Malformations: Clinical Features and Molecular Characterization of Novel Variants. Journal of Molecular Neuroscience, 2021, 71, 1876-1883.	2.3	8
4	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	9.0	46
5	A Novel Variant in Superoxide Dismutase 1 Gene (p.V119M) in Als Patients with Pure Lower Motor Neuron Presentation. Genes, 2021, 12, 1544.	2.4	3
6	Concern regarding classification of c.703G>A/p.Gly235Arg as a novel missense variant in KRIT1 gene. Human Mutation, 2020, 41, 1069-1071.	2.5	1
7	Skeletal-Muscle Metabolic Reprogramming in ALS-SOD1G93A Mice Predates Disease Onset and Is A Promising Therapeutic Target. IScience, 2020, 23, 101087.	4.1	55
8	Genotypeâ€“phenotype correlation and evidence for a common ancestor in two Italian ALS patients with the D124G SOD1 mutation. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2019, 20, 611-614.	1.7	3
9	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. Annals of Neurology, 2019, 85, 470-481.	5.3	118
10	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	8.1	517
11	MicroRNAs as Biomarkers in Amyotrophic Lateral Sclerosis. Cells, 2018, 7, 219.	4.1	74
12	Impaired intracortical transmission in G2019S leucine richâ€repeat kinase Parkinson patients. Movement Disorders, 2017, 32, 750-756.	3.9	16
13	C9ORF72 gene expansion in a patient with intellectual disability and psychiatric disease. Neurological Sciences, 2017, 38, 207-208.	1.9	0
14	TBK1 is associated with ALS and ALS-FTD in Sardinian patients. Neurobiology of Aging, 2016, 43, 180.e1-180.e5.	3.1	40
15	Identification of miRNAs as Potential Biomarkers in Cerebrospinal Fluid from Amyotrophic Lateral Sclerosis Patients. NeuroMolecular Medicine, 2016, 18, 551-560.	3.4	67
16	ATNX2 is not a regulatory gene in Italian amyotrophic lateral sclerosis patients with C9ORF72 GGGGCC expansion. Neurobiology of Aging, 2016, 39, 218.e5-218.e8.	3.1	6
17	Lack of relationship between the P413L chromogranin B variant and a SALS Italian cohort. Gene, 2015, 568, 186-189.	2.2	4
18	HFE p.H63D polymorphism does not influence ALS phenotype and survival. Neurobiology of Aging, 2015, 36, 2906.e7-2906.e11.	3.1	8

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19	Erythropoietin in amyotrophic lateral sclerosis: a multicentre, randomised, double blind, placebo controlled, phase III study. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 879-886.	1.9	32
20	CHCH10 mutations in an Italian cohort of familial and sporadic amyotrophic lateral sclerosis patients. Neurobiology of Aging, 2015, 36, 1767.e3-1767.e6.	3.1	44
21	ATXN2 is a modifier of phenotype in ALS patients of Sardinian ancestry. Neurobiology of Aging, 2015, 36, 2906.e1-2906.e5.	3.1	19
22	Mutations in the Matrin 3 gene cause familial amyotrophic lateral sclerosis. Nature Neuroscience, 2014, 17, 664-666.	14.8	398
23	Genetic counselling in ALS: facts, uncertainties and clinical suggestions. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 478-485.	1.9	99
24	Genetic architecture of ALS in Sardinia. Neurobiology of Aging, 2014, 35, 2882.e7-2882.e12.	3.1	60
25	Double trouble? Progranulin mutation and C9ORF72 repeat expansion in a case of primary non-fluent aphasia. Journal of the Neurological Sciences, 2014, 341, 176-178.	0.6	17
26	Genotyping of Macrophage Migration Inhibitory Factor (MIF) CATT5â€“8 Repeat Polymorphism by Denaturing High-Performance Liquid Chromatography (DHPLC). Molecular Biotechnology, 2013, 54, 874-879.	2.4	1
27	Recurrent G41S mutation in Cu/Zn superoxide dismutase gene (SOD1) causing familial amyotrophic lateral sclerosis in a large Polish family. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 132-136.	2.1	5
28	Replication of association of CHRNA4 rare variants with sporadic amyotrophic lateral sclerosis: The Italian multicentre study. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 580-584.	2.1	7
29	No association of MTHFR c.677C>T variant with sporadic ALS in an Italian population. Neurobiology of Aging, 2012, 33, 208.e7-208.e8.	3.1	4
30	C9ORF72 hexanucleotide repeat expansions in the Italian sporadic ALS population. Neurobiology of Aging, 2012, 33, 1848.e15-1848.e20.	3.1	76
31	Clinical characteristics of patients with familial amyotrophic lateral sclerosis carrying the pathogenic GGGGCC hexanucleotide repeat expansion of C9ORF72. Brain, 2012, 135, 784-793.	7.6	182
32	Frequency of the C9orf72 hexanucleotide repeat expansion in patients with amyotrophic lateral sclerosis and frontotemporal dementia: a cross-sectional study. Lancet Neurology, The, 2012, 11, 323-330.	10.2	1,039
33	FUS mutations in sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2011, 32, 550.e1-550.e4.	3.1	79
34	Lack of association of PON polymorphisms with sporadic ALS in an Italian population. Neurobiology of Aging, 2011, 32, 552.e7-552.e13.	3.1	18
35	Exome Sequencing Reveals VCP Mutations as a Cause of Familial ALS. Neuron, 2011, 69, 397.	8.1	7
36	A Hexanucleotide Repeat Expansion in C9ORF72 Is the Cause of Chromosome 9p21-Linked ALS-FTD. Neuron, 2011, 72, 257-268.	8.1	3,833

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37	Regulation of redox forms of plasma thiols by albumin in multiple sclerosis after fasting and methionine loading test. Amino Acids, 2010, 38, 1461-1471.	2.7	24
38	Genetic Variations Within KRIT1/CCM1, MGC4607/CCM2 and PDCD10/CCM3 in a Large Italian Family Harbouring a Krit1/CCM1 Mutation. Journal of Molecular Neuroscience, 2010, 42, 235-242.	2.3	18
39	Cavernous Malformation of the Optic Nerve Mimicking Optic Neuritis. Journal of Neuro-Ophthalmology, 2010, 30, 126-131.	0.8	18
40	Severe familial ALS with a novel exon 4 mutation (L106F) in the SOD1 gene. Journal of the Neurological Sciences, 2010, 293, 112-115.	0.6	17
41	Exome Sequencing Reveals VCP Mutations as a Cause of Familial ALS. Neuron, 2010, 68, 857-864.	8.1	1,100
42	G41SSOD1mutation: A common ancestor for six ALS Italian families with an aggressive phenotype. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2010, 11, 210-215.	2.1	14
43	A novel exon 1 mutation (G10R) in theSOD1gene in a patient with familial ALS. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2010, 11, 481-485.	2.1	2
44	D90A-SOD1 mutation in ALS: The first report of heterozygous Italian patients and unusual findings. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2010, 11, 216-219.	2.1	24
45	A two-stage genome-wide association study of sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2009, 18, 1524-1532.	2.9	106
46	CACNA1A gene non-synonymous single nucleotide polymorphisms and common migraine in Italy: a case-control association study with a micro-array technology. Clinical Chemistry and Laboratory Medicine, 2009, 47, 783-5.	2.3	5
47	Two Italian kindreds with familial amyotrophic lateral sclerosis due to FUS mutation. Neurobiology of Aging, 2009, 30, 1272-1275.	3.1	128
48	Clinical, Magnetic Resonance Imaging, and Genetic Study of 5 Italian Families With Cerebral Cavernous Malformation. Archives of Neurology, 2007, 64, 843.	4.5	37
49	Variations in the coding and regulatory sequences of the angiogenin (ANG) gene are not associated to ALS (amyotrophic lateral sclerosis) in the Italian population. Journal of the Neurological Sciences, 2007, 258, 123-127.	0.6	37
50	SOD1 mutations in amyotrophic lateral sclerosis. Journal of Neurology, 2005, 252, 782-788.	3.6	86
51	Activity of protein phosphatase calcineurin is decreased in sporadic and familial amyotrophic lateral sclerosispatients. Journal of Neurochemistry, 2004, 90, 1237-1242.	3.9	34
52	Wilson?s disease with Leu492Ser mutation and arylsulfatase A pseudodeficiency: just a coincidence?. Neurological Sciences, 2004, 25, 18-20.	1.9	0
53	Posterior knee pain: primary symptom of a small non-occlusive venous clot. Archives of Disease in Childhood, 2003, 88, 728-729.	1.9	0
54	Familial hemiplegic migraine: a ion channel disorder. Brain Research Bulletin, 2001, 56, 239-241.	3.0	30

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55	Deep Vein Thrombosis during Varicella in a Child with Factor V Leiden Mutation and Familial Deficiency of Protein S. Thrombosis and Haemostasis, 2001, 85, 370-370.	3.4	0
56	Molecular basis of late-life globoid cell leukodystrophy. Human Mutation, 1999, 14, 256-262.	2.5	23
57	Potential Role of 11?-Hydroxysteroid Dehydrogenase in Human Trophoblast-Endometrial Interactions,b. Annals of the New York Academy of Sciences, 1996, 784, 433-438.	3.8	5
58	A Human Kidney cDNA Which Induces a Cell Surface Protein Epitope Recognized by a Monoclonal Antibody against Galactosylceramide. Biochemical and Biophysical Research Communications, 1996, 227, 636-641.	2.1	0
59	Late-onset G sub M2 gangliosidosis. Neurology, 1996, 47, 547-552.	1.1	22
60	Substitution of Alanine543 with a Threonine Residue at the Carboxy Terminal End of the Î²-Chain Is Associated with Thermolabile Hexosaminidase B in a Jewish Family of Oriental Ancestry. Biochemical and Molecular Medicine, 1995, 56, 31-36.	1.4	7
61	Cherry-Red Spot Myoclonus Syndrome (Type I Sialidosis). Developmental Neuroscience, 1991, 13, 320-326.	2.0	29