

Stefania Battistini

List of Publications by Year in descending order

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papers

8,681
citations

218381

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docs citations

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times ranked

9350
citing authors

#	ARTICLE	IF	CITATIONS
1	A Hexanucleotide Repeat Expansion in C9ORF72 Is the Cause of Chromosome 9p21-Linked ALS-FTD. <i>Neuron</i> , 2011, 72, 257-268.	3.8	3,833
2	Exome Sequencing Reveals VCP Mutations as a Cause of Familial ALS. <i>Neuron</i> , 2010, 68, 857-864.	3.8	1,100
3	Frequency of the C9orf72 hexanucleotide repeat expansion in patients with amyotrophic lateral sclerosis and frontotemporal dementia: a cross-sectional study. <i>Lancet Neurology</i> , The, 2012, 11, 323-330.	4.9	1,039
4	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , 2018, 97, 1268-1283.e6.	3.8	517
5	Mutations in the Matrin 3 gene cause familial amyotrophic lateral sclerosis. <i>Nature Neuroscience</i> , 2014, 17, 664-666.	7.1	398
6	Clinical characteristics of patients with familial amyotrophic lateral sclerosis carrying the pathogenic GGGGCC hexanucleotide repeat expansion of C9ORF72. <i>Brain</i> , 2012, 135, 784-793.	3.7	182
7	Two Italian kindreds with familial amyotrophic lateral sclerosis due to FUS mutation. <i>Neurobiology of Aging</i> , 2009, 30, 1272-1275.	1.5	128
8	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 2019, 85, 470-481.	2.8	118
9	A two-stage genome-wide association study of sporadic amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2009, 18, 1524-1532.	1.4	106
10	Genetic counselling in ALS: facts, uncertainties and clinical suggestions. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, 478-485.	0.9	99
11	SOD1 mutations in amyotrophic lateral sclerosis. <i>Journal of Neurology</i> , 2005, 252, 782-788.	1.8	86
12	FUS mutations in sporadic amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2011, 32, 550.e1-550.e4.	1.5	79
13	C9ORF72 hexanucleotide repeat expansions in the Italian sporadic ALS population. <i>Neurobiology of Aging</i> , 2012, 33, 1848.e15-1848.e20.	1.5	76
14	MicroRNAs as Biomarkers in Amyotrophic Lateral Sclerosis. <i>Cells</i> , 2018, 7, 219.	1.8	74
15	Identification of miRNAs as Potential Biomarkers in Cerebrospinal Fluid from Amyotrophic Lateral Sclerosis Patients. <i>NeuroMolecular Medicine</i> , 2016, 18, 551-560.	1.8	67
16	Genetic architecture of ALS in Sardinia. <i>Neurobiology of Aging</i> , 2014, 35, 2882.e7-2882.e12.	1.5	60
17	Skeletal-Muscle Metabolic Reprogramming in ALS-SOD1G93A Mice Predates Disease Onset and Is A Promising Therapeutic Target. <i>IScience</i> , 2020, 23, 101087.	1.9	55
18	Association of Variants in the <i>SPTLC1</i> Gene With Juvenile Amyotrophic Lateral Sclerosis. <i>JAMA Neurology</i> , 2021, 78, 1236.	4.5	46

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19	CHCH10 mutations in an Italian cohort of familial and sporadic amyotrophic lateral sclerosis patients. <i>Neurobiology of Aging</i> , 2015, 36, 1767.e3-1767.e6.	1.5	44
20	TBK1 is associated with ALS and ALS-FTD in Sardinian patients. <i>Neurobiology of Aging</i> , 2016, 43, 180.e1-180.e5.	1.5	40
21	Clinical, Magnetic Resonance Imaging, and Genetic Study of 5 Italian Families With Cerebral Cavernous Malformation. <i>Archives of Neurology</i> , 2007, 64, 843.	4.9	37
22	Variations in the coding and regulatory sequences of the angiogenin (ANG) gene are not associated to ALS (amyotrophic lateral sclerosis) in the Italian population. <i>Journal of the Neurological Sciences</i> , 2007, 258, 123-127.	0.3	37
23	Activity of protein phosphatase calcineurin is decreased in sporadic and familial amyotrophic lateral sclerosis patients. <i>Journal of Neurochemistry</i> , 2004, 90, 1237-1242.	2.1	34
24	Erythropoietin in amyotrophic lateral sclerosis: a multicentre, randomised, double blind, placebo controlled, phase III study. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2015, 86, 879-886.	0.9	32
25	Familial hemiplegic migraine: a ion channel disorder. <i>Brain Research Bulletin</i> , 2001, 56, 239-241.	1.4	30
26	Cherry-Red Spot Myoclonus Syndrome (Type I Sialidosis). <i>Developmental Neuroscience</i> , 1991, 13, 320-326.	1.0	29
27	Regulation of redox forms of plasma thiols by albumin in multiple sclerosis after fasting and methionine loading test. <i>Amino Acids</i> , 2010, 38, 1461-1471.	1.2	24
28	D90A-SOD1 mutation in ALS: The first report of heterozygous Italian patients and unusual findings. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2010, 11, 216-219.	2.3	24
29	Molecular basis of late-life globoid cell leukodystrophy. , 1999, 14, 256-262.		23
30	Late-onset G sub M2 gangliosidosis. <i>Neurology</i> , 1996, 47, 547-552.	1.5	22
31	Molecular Genetic Features of Cerebral Cavernous Malformations (CCM) Patients: An Overall View from Genes to Endothelial Cells. <i>Cells</i> , 2021, 10, 704.	1.8	21
32	ATXN2 is a modifier of phenotype in ALS patients of Sardinian ancestry. <i>Neurobiology of Aging</i> , 2015, 36, 2906.e1-2906.e5.	1.5	19
33	Genetic Variations Within KRIT1/CCM1, MGC4607/CCM2 and PDCD10/CCM3 in a Large Italian Family Harboring a Krit1/CCM1 Mutation. <i>Journal of Molecular Neuroscience</i> , 2010, 42, 235-242.	1.1	18
34	Cavernous Malformation of the Optic Nerve Mimicking Optic Neuritis. <i>Journal of Neuro-Ophthalmology</i> , 2010, 30, 126-131.	0.4	18
35	Lack of association of PON polymorphisms with sporadic ALS in an Italian population. <i>Neurobiology of Aging</i> , 2011, 32, 552.e7-552.e13.	1.5	18
36	Severe familial ALS with a novel exon 4 mutation (L106F) in the SOD1 gene. <i>Journal of the Neurological Sciences</i> , 2010, 293, 112-115.	0.3	17

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37	Double trouble? Progranulin mutation and C9ORF72 repeat expansion in a case of primary non-fluent aphasia. <i>Journal of the Neurological Sciences</i> , 2014, 341, 176-178.	0.3	17
38	Impaired intracortical transmission in G2019S leucine rich repeat kinase Parkinson patients. <i>Movement Disorders</i> , 2017, 32, 750-756.	2.2	16
39	G41S SOD1 mutation: A common ancestor for six ALS Italian families with an aggressive phenotype. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2010, 11, 210-215.	2.3	14
40	HFE p.H63D polymorphism does not influence ALS phenotype and survival. <i>Neurobiology of Aging</i> , 2015, 36, 2906.e7-2906.e11.	1.5	8
41	KRIT1 Gene in Patients with Cerebral Cavernous Malformations: Clinical Features and Molecular Characterization of Novel Variants. <i>Journal of Molecular Neuroscience</i> , 2021, 71, 1876-1883.	1.1	8
42	BDNF and Pro-BDNF in Amyotrophic Lateral Sclerosis: A New Perspective for Biomarkers of Neurodegeneration. <i>Brain Sciences</i> , 2022, 12, 617.	1.1	8
43	Substitution of Alanine543 with a Threonine Residue at the Carboxy Terminal End of the \hat{I}^2 -Chain Is Associated with Thermolabile Hexosaminidase B in a Jewish Family of Oriental Ancestry. <i>Biochemical and Molecular Medicine</i> , 1995, 56, 31-36.	1.5	7
44	Exome Sequencing Reveals VCP Mutations as a Cause of Familial ALS. <i>Neuron</i> , 2011, 69, 397.	3.8	7
45	Replication of association of CHRNA4 rare variants with sporadic amyotrophic lateral sclerosis: The Italian multicentre study. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2012, 13, 580-584.	2.3	7
46	ATNX2 is not a regulatory gene in Italian amyotrophic lateral sclerosis patients with C9ORF72 GGGGCC expansion. <i>Neurobiology of Aging</i> , 2016, 39, 218.e5-218.e8.	1.5	6
47	Potential Role of 11 β -Hydroxysteroid Dehydrogenase in Human Trophoblast-Endometrial Interactions. <i>Annals of the New York Academy of Sciences</i> , 1996, 784, 433-438.	1.8	5
48	CACNA1A gene non-synonymous single nucleotide polymorphisms and common migraine in Italy: a case-control association study with a micro-array technology. <i>Clinical Chemistry and Laboratory Medicine</i> , 2009, 47, 783-5.	1.4	5
49	Recurrent G41S mutation in Cu/Zn superoxide dismutase gene (SOD1) causing familial amyotrophic lateral sclerosis in a large Polish family. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2012, 13, 132-136.	2.3	5
50	No association of MTHFR c.677C>T variant with sporadic ALS in an Italian population. <i>Neurobiology of Aging</i> , 2012, 33, 208.e7-208.e8.	1.5	4
51	Lack of relationship between the P413L chromogranin B variant and a SALS Italian cohort. <i>Gene</i> , 2015, 568, 186-189.	1.0	4
52	Genotype-phenotype correlation and evidence for a common ancestor in two Italian ALS patients with the D124G SOD1 mutation. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2019, 20, 611-614.	1.1	3
53	A Novel Variant in Superoxide Dismutase 1 Gene (p.V119M) in Als Patients with Pure Lower Motor Neuron Presentation. <i>Genes</i> , 2021, 12, 1544.	1.0	3
54	A novel exon 1 mutation (G10R) in the SOD1 gene in a patient with familial ALS. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2010, 11, 481-485.	2.3	2

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55	Genotyping of Macrophage Migration Inhibitory Factor (MIF) CATT5â€8 Repeat Polymorphism by Denaturing High-Performance Liquid Chromatography (DHPLC). <i>Molecular Biotechnology</i> , 2013, 54, 874-879.	1.3	1
56	Concern regarding classification of c.703G>A/p.Gly235Arg as a novel missense variant in KRIT1 gene. <i>Human Mutation</i> , 2020, 41, 1069-1071.	1.1	1
57	A Human Kidney cDNA Which Induces a Cell Surface Protein Epitope Recognized by a Monoclonal Antibody against Galactosylceramide. <i>Biochemical and Biophysical Research Communications</i> , 1996, 227, 636-641.	1.0	0
58	Deep Vein Thrombosis during Varicella in a Child with Factor V Leiden Mutation and Familial Deficiency of Protein S. <i>Thrombosis and Haemostasis</i> , 2001, 85, 370-370.	1.8	0
59	Posterior knee pain: primary symptom of a small non-occlusive venous clot. <i>Archives of Disease in Childhood</i> , 2003, 88, 728-729.	1.0	0
60	Wilson's disease with Leu492Ser mutation and arylsulfatase A pseudodeficiency: just a coincidence?. <i>Neurological Sciences</i> , 2004, 25, 18-20.	0.9	0
61	C9ORF72 gene expansion in a patient with intellectual disability and psychiatric disease. <i>Neurological Sciences</i> , 2017, 38, 207-208.	0.9	0