Gianni SorarÃ¹

List of Publications by Year
in descending order

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107 papers	5,443 citations	109321 35 h-index	98798 67 g-index
110 all docs	110 docs citations	110 times ranked	7837 citing authors

#	Article	IF	Citations
1	Quality of life assessment in adult spinal muscular atrophy patients treated with nusinersen. Journal of Neurology, 2022, 269, 3264-3275.	3.6	6
2	The relevance of migraine in the clinical spectrum of mitochondrial disorders. Scientific Reports, 2022, 12, 4222.	3.3	7
3	AR cooperates with SMAD4 to maintain skeletal muscle homeostasis. Acta Neuropathologica, 2022, 143, 713-731.	7.7	6
4	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 448-460.e4.	8.1	56
5	The unfolded protein response in amyotrophic later sclerosis: results of a phase 2 trial. Brain, 2021, 144, 2635-2647.	7.6	33
6	Evaluation of peripherin in biofluids of patients with motor neuron diseases. Annals of Clinical and Translational Neurology, 2021, 8, 1750-1754.	3.7	11
7	Decoding distinctive features of plasma extracellular vesicles in amyotrophic lateral sclerosis. Molecular Neurodegeneration, 2021, 16, 52.	10.8	19
8	Brainstem glucose hypermetabolism in ALS/FTD and shorten survival: a ¹⁸F-FDG PET/MR study. Journal of Nuclear Medicine, 2021, , jnumed.121.262232.	5.0	9
9	Empathy-based supportive treatment in amyotrophic lateral sclerosis: A pragmatic study. American Journal of Clinical Hypnosis, 2021, 63, 202-216.	0.6	7
10	Enhanced Neural Empathic Responses in Patients with Spino-Bulbar Muscular Atrophy: An Electrophysiological Study. Brain Sciences, 2021, 11, 16.	2.3	7
11	CSF Heavy Neurofilament May Discriminate and Predict Motor Neuron Diseases with Upper Motor Neuron Involvement. Biomedicines, 2021, 9, 1623.	3.2	4
12	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. Nature Genetics, 2021, 53, 1636-1648.	21.4	223
13	The Role of Motor System in Mental Rotation: New Insights from Myotonic Dystrophy Type 1. Journal of the International Neuropsychological Society, 2020, 26, 492-502.	1.8	3
14	Neurofilaments in motor neuron disorders: towards promising diagnostic and prognostic biomarkers. Molecular Neurodegeneration, 2020, 15, 58.	10.8	68
15	Clinical features and outcomes of the flail arm and flail leg and pure lower motor neuron MND variants: a multicentre Italian study. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 1001-1003.	1.9	14
16	FUS ALS-causative mutations impair FUS autoregulation and splicing factor networks through intron retention. Nucleic Acids Research, 2020, 48, 6889-6905.	14.5	70
17	The Italian multicenter experience with edaravone in amyotrophic lateral sclerosis. Journal of Neurology, 2020, 267, 3258-3267.	3.6	37
18	MetabolicÂalterations in spinal and bulbar muscular atrophy. Revue Neurologique, 2020, 176, 780-787.	1.5	7

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19	Sorting Rare ALS Genetic Variants by Targeted Re-Sequencing Panel in Italian Patients: OPTN, VCP, and SQSTM1 Variants Account for 3% of Rare Genetic Forms. Journal of Clinical Medicine, 2020, 9, 412.	2.4	24
20	MEF2 impairment underlies skeletal muscle atrophy in polyglutamine disease. Acta Neuropathologica, 2020, 140, 63-80.	7.7	23
21	Polyglutamine-Expanded Androgen Receptor Alteration of Skeletal Muscle Homeostasis and Myonuclear Aggregation Are Affected by Sex, Age and Muscle Metabolism. Cells, 2020, 9, 325.	4.1	21
22	Transforming growth factor beta 1 signaling is altered in the spinal cord and muscle of amyotrophic lateral sclerosis mice and patients. Neurobiology of Aging, 2019, 82, 48-59.	3.1	15
23	Safety and efficacy of edaravone compared to historical controls in patients with amyotrophic lateral sclerosis from North-Eastern Italy. Journal of the Neurological Sciences, 2019, 404, 47-51.	0.6	16
24	Comparative Analysis of C9orf72 and Sporadic Disease in a Large Multicenter ALS Population: The Effect of Male Sex on Survival of C9orf72 Positive Patients. Frontiers in Neuroscience, 2019, 13, 485.	2.8	35
25	Proteostasis and ALS: protocol for a phase II, randomised, double-blind, placebo-controlled, multicentre clinical trial for colchicine in ALS (Co-ALS). BMJ Open, 2019, 9, e028486.	1.9	44
26	Results from Phase I Clinical Trial with Intraspinal Injection of Neural Stem Cells in Amyotrophic Lateral Sclerosis: A Long-Term Outcome. Stem Cells Translational Medicine, 2019, 8, 887-897.	3.3	71
27	Collagen XIX Alpha 1 Improves Prognosis in Amyotrophic Lateral Sclerosis. , 2019, 10, 278.		18
28	Preliminary design and validation of the â€œ6-K-scaleâ€•for bulbar symptoms evaluation in SBMA. Neurological Sciences, 2019, 40, 1393-1401.	1.9	2
29	Brain MRI shows white matter sparing in Kennedy's disease and slowâ€progressing lower motor neuron disease. Human Brain Mapping, 2019, 40, 3102-3112.	3.6	12
30	Muscle and not neuronal biomarkers correlate with severity in spinal and bulbar muscular atrophy. Neurology, 2019, 92, e1205-e1211.	1.1	41
31	Safety and efficacy of nabiximols on spasticity symptoms in patients with motor neuron disease (CANALS): a multicentre, double-blind, randomised, placebo-controlled, phase 2 trial. Lancet Neurology, The, 2019, 18, 155-164.	10.2	63
32	Longitudinal evaluation of SMN levels as biomarker for spinal muscular atrophy: results of a phase IIb double-blind study of salbutamol. Journal of Medical Genetics, 2019, 56, 293-300.	3.2	30
33	Insights into the genetic epidemiology of spinal and bulbar muscular atrophy: prevalence estimation and multiple founder haplotypes in the Veneto Italian region. European Journal of Neurology, 2019, 26, 519-524.	3.3	12
34	New <scp>FIG</scp>4 gene mutations causing aggressive <scp>ALS</scp>. European Journal of Neurology, 2018, 25, e41-e42.	3.3	14
35	Beyond motor neurons: expanding the clinical spectrum in Kennedyâ€™s disease. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 808-812.	1.9	48
36	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	8.1	517

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37	Rapamycin treatment for amyotrophic lateral sclerosis. Medicine (United States), 2018, 97, e11119.	1.0	96
38	The clinical spectrum of CASQ1-related myopathy. Neurology, 2018, 91, e1629-e1641.	1.1	14
39	Safety, tolerability, and preliminary efficacy of an IGF-1 mimetic in patients with spinal and bulbar muscular atrophy: a randomised, placebo-controlled trial. Lancet Neurology, The, 2018, 17, 1043-1052.	10.2	28
40	Unimpaired Neuropsychological Performance and Enhanced Memory Recall in Patients with Sbma: A Large Sample Comparative Study. Scientific Reports, 2018, 8, 13627.	3.3	8
41	ATXN2 trinucleotide repeat length correlates with risk of ALS. Neurobiology of Aging, 2017, 51, 178.e1-178.e9.	3.1	86
42	Increased mitophagy in the skeletal muscle of spinal and bulbar muscular atrophy patients. Human Molecular Genetics, 2017, 26, ddx019.	2.9	37
43	Beta-agonist stimulation ameliorates the phenotype of spinal and bulbar muscular atrophy mice and patient-derived myotubes. Scientific Reports, 2017, 7, 41046.	3.3	26
44	Diagnostic and Prognostic Biomarkers in Amyotrophic Lateral Sclerosis. JAMA Neurology, 2017, 74, 525.	9.0	139
45	SPP1 genotype and glucocorticoid treatment modify osteopontin expression in Duchenne muscular dystrophy cells. Human Molecular Genetics, 2017, 26, 3342-3351.	2.9	23
46	Humanized mutant FUS drives progressive motor neuron degeneration without aggregation in â€˜FUSDelta14â€™ knockin mice. Brain, 2017, 140, 2797-2805.	7.6	95
47	TBK1mutations in Italian patients with amyotrophic lateral sclerosis: genetic and functional characterisation. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 869-875.	1.9	38
48	Comorbidity of dementia with amyotrophic lateral sclerosis (ALS): insights from a large multicenter Italian cohort. Journal of Neurology, 2017, 264, 2224-2231.	3.6	19
49	Multimodal structural MRI in the diagnosis of motor neuron diseases. NeuroImage: Clinical, 2017, 16, 240-247.	2.7	55
50	Protein misfolding, amyotrophic lateral sclerosis and guanabenz: protocol for a phase II RCT with futility design (ProMISe trial). BMJ Open, 2017, 7, e015434.	1.9	14
51	Muscle MRI and functional outcome measures in Becker muscular dystrophy. Scientific Reports, 2017, 7, 16060.	3.3	35
52	Factors predicting survival in ALS: a multicenter Italian study. Journal of Neurology, 2017, 264, 54-63.	3.6	96
53	No effect of <scp>AR</scp> polyG polymorphism on spinal and bulbar muscular atrophy phenotype. European Journal of Neurology, 2016, 23, 1134-1136.	3.3	8
54	Mitochondrial implications in bulbospinal muscular atrophy (Kennedy disease). Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2016, 17, 112-118.	1.7	6

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55	Intraspinal stem cell transplantation for amyotrophic lateral sclerosis: Ready for efficacy clinical trials?. Cytotherapy, 2016, 18, 1471-1475.	0.7	21
56	Validation of the Italian version of the SBMA Functional Rating Scale as outcome measure. Neurological Sciences, 2016, 37, 1815-1821.	1.9	9
57	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1043-1048.	21.4	494
58	Functional changes in Becker muscular dystrophy: implications for clinical trials in dystrophinopathies. Scientific Reports, 2016, 6, 32439.	3.3	36
59	Association of a Locus in theCAMTA1Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. JAMA Neurology, 2016, 73, 812.	9.0	57
60	Towards a European Registry and Biorepository for Patients with Spinal and Bulbar Muscular Atrophy. Journal of Molecular Neuroscience, 2016, 58, 394-400.	2.3	10
61	Non-neural phenotype of spinal and bulbar muscular atrophy: results from a large cohort of Italian patients. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 810-816.	1.9	59
62	Glycolytic-to-oxidative fiber-type switch and mTOR signaling activation are early-onset features of SBMA muscle modified by high-fat diet. Acta Neuropathologica, 2016, 132, 127-144.	7.7	74
63	Clinical and molecular study in a long-surviving patient with MLASA syndrome due to novel PUS1 mutations. Neurogenetics, 2016, 17, 65-70.	1.4	29
64	Onset Manifestations of Spinal and Bulbar Muscular Atrophy (Kennedyâ€™s Disease). Journal of Molecular Neuroscience, 2016, 58, 321-329.	2.3	40
65	Hypnosis-based psychodynamic treatment in ALS: a longitudinal study on patients and their caregivers. Frontiers in Psychology, 2015, 6, 822.	2.1	29
66	210th ENMC International Workshop: Research and clinical management of patients with spinal and bulbar muscular atrophy, 27â€“29 March, 2015, Naarden, The Netherlands. Neuromuscular Disorders, 2015, 25, 802-812.	0.6	16
67	Theory of mind, empathy and neuropsychological functioning in X-linked Spinal and Bulbar Muscular Atrophy: a controlled study of 20 patients. Journal of Neurology, 2015, 262, 394-401.	3.6	21
68	Cardiac Function in Types II and III Spinal Muscular Atrophy: Should We Change Standards of Care?. Neuropediatrics, 2015, 46, 033-036.	0.6	9
69	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
70	Female gender doubles executive dysfunction risk in ALS: a case-control study in 165 patients. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 574-579.	1.9	26
71	Impact on children of a parent with ALS: a case-control study. Frontiers in Psychology, 2015, 6, 288.	2.1	16
72	TUBA4A gene analysis in sporadic amyotrophic lateral sclerosis: identification of novel mutations. Journal of Neurology, 2015, 262, 1376-1378.	3.6	44

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73	Human neural stem cell transplantation in ALS: initial results from a phase I trial. Journal of Translational Medicine, 2015, 13, 17.	4.4	151
74	Genetic Modifiers of Duchenne Muscular Dystrophy and Dilated Cardiomyopathy. PLoS ONE, 2015, 10, e0141240.	2.5	58
75	Disease Propagation in Amyotrophic Lateral Sclerosis: Insights and Hopes. Journal of Neurology & Stroke, 2015, 2, .	0.1	0
76	The blurred scenario of motor neuron disorders linked toSpatacsinmutations: a case report. European Journal of Neurology, 2014, 21, e85-e86.	3.3	2
77	Improving the knowledge of amyotrophic lateral sclerosis genetics: novel SOD1 and FUS variants. Neurobiology of Aging, 2014, 35, 1212.e7-1212.e10.	3.1	13
78	A genome-wide association meta-analysis identifies a novel locus at 17q11.2 associated with sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2014, 23, 2220-2231.	2.9	123
79	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. Neuron, 2014, 84, 324-331.	8.1	308
80	Skeletal Muscle Satellite Cells in Amyotrophic Lateral Sclerosis. Ultrastructural Pathology, 2014, 38, 295-302.	0.9	37
81	Androgen-dependent impairment of myogenesis in spinal and bulbar muscular atrophy. Acta Neuropathologica, 2013, 126, 109-121.	7.7	41
82	Screening of the PFN1 gene in sporadic amyotrophic lateral sclerosis and in frontotemporal dementia. Neurobiology of Aging, 2013, 34, 1517.e9-1517.e10.	3.1	35
83	Analysis of hnRNPA1, A2/B1, and A3 genes in patients with amyotrophic lateral sclerosis. Neurobiology of Aging, 2013, 34, 2695.e11-2695.e12.	3.1	30
84	Disruption of skeletal muscle mitochondrial network genes and miRNAs in amyotrophic lateral sclerosis. Neurobiology of Disease, 2013, 49, 107-117.	4.4	194
85	Clinical and molecular cross-sectional study of a cohort of adult type III spinal muscular atrophy patients: clues from a biomarker study. European Journal of Human Genetics, 2013, 21, 630-636.	2.8	39
86	Pilot trial of clenbuterol in spinal and bulbar muscular atrophy. Neurology, 2013, 80, 2095-2098.	1.1	47
87	Specific numerical processing impairment in ALS patients. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 6-12.	1.7	8
88	Ubiquilin 2mutations in Italian patients with amyotrophic lateral sclerosis and frontotemporal dementia. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 183-187.	1.9	74
89	No evidence of cardiomyopathy in spinal and bulbar muscular atrophy. Acta Neurologica Scandinavica, 2013, 128, e30-e32.	2.1	14
90	Cardiomyopathy in patients with POMT1-related congenital and limb-girdle muscular dystrophy. European Journal of Human Genetics, 2012, 20, 1234-1239.	2.8	31

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91	Parkinson-like features in ALS with predominant upper motor neuron involvement. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 137-143.	2.1	18
92	TGFBR2 but not SPP1 genotype modulates osteopontin expression in Duchenne muscular dystrophy muscle. Journal of Pathology, 2012, 228, 251-259.	4.5	22
93	Mutational analysis of VCP gene in familial amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 630.e1-630.e2.	3.1	17
94	C9ORF72 repeat expansion in a large Italian ALS cohort: evidence of a founder effect. Neurobiology of Aging, 2012, 33, 2528.e7-2528.e14.	3.1	74
95	Efficacy of Hypnosis-Based Treatment in Amyotrophic Lateral Sclerosis: A Pilot Study. Frontiers in Psychology, 2012, 3, 465.	2.1	19
96	No association of DPP6 with amyotrophic lateral sclerosis in an Italian population. Neurobiology of Aging, 2011, 32, 966-967.	3.1	28
97	Clinical and molecular characterization of limbâ€girdle muscular dystrophy due to LAMA2 mutations. Muscle and Nerve, 2011, 44, 703-709.	2.2	52
98	ALS risk but not phenotype is affected by ataxin-2 intermediate length polyglutamine expansion. Neurology, 2011, 76, 2030-2031.	1.1	25
99	Psychopathological features and suicidal ideation in amyotrophic lateral sclerosis patients. Neurological Sciences, 2010, 31, 735-740.	1.9	22
100	Natural history of upper motor neuron-dominant ALS. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2010, 11, 424-429.	2.1	29
101	TDP-43 in skeletal muscle of patients affected with amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2010, 11, 240-243.	2.1	19
102	Mutations of FUS gene in sporadic amyotrophic lateral sclerosis. Journal of Medical Genetics, 2010, 47, 190-194.	3.2	152
103	Progress in enzyme replacement therapy in glycogen storage disease type II. Therapeutic Advances in Neurological Disorders, 2009, 2, 143-153.	3.5	30
104	Spinal and bulbar muscular atrophy: Skeletal muscle pathology in male patients and heterozygous females. Journal of the Neurological Sciences, 2008, 264, 100-105.	0.6	133
105	Muscle histopathology in upper motor neuron-dominant amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2008, 9, 287-293.	2.1	13
106	A pilot trial with clenbuterol in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2006, 7, 252-254.	2.1	14
107	Human skeletal muscle atrophy in amyotrophic lateral sclerosis reveals a reduction in Akt and an increase in atroginâ€1. FASEB Journal, 2006, 20, 583-585.	0.5	127