## Nicola Ticozzi

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	A preliminary comparison between ECAS and ALS-CBS in classifying cognitive–behavioural phenotypes in a cohort of non-demented amyotrophic lateral sclerosis patients. Journal of Neurology, 2022, 269, 1899-1904.	1.8	5
2	Genome-wide identification of the genetic basis of amyotrophic lateral sclerosis. Neuron, 2022, 110, 992-1008.e11.	3.8	51
3	Upper motor neuron dysfunction is associated with the presence of behavioural impairment in patients with amyotrophic lateral sclerosis. European Journal of Neurology, 2022, 29, 1402-1409.	1.7	9
4	Genome-wide study of DNA methylation shows alterations in metabolic, inflammatory, and cholesterol pathways in ALS. Science Translational Medicine, 2022, 14, eabj0264.	5.8	38
5	Gaze-Contingent Eye-Tracking Training in Brain Disorders: A Systematic Review. Brain Sciences, 2022, 12, 931.	1.1	6
6	Cerebrospinal fluid phosphorylated neurofilament heavy chain and chitotriosidase in primary lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 221-223.	0.9	9
7	The Effect of <scp><i>SMN</i></scp> Gene Dosage on <scp>ALS</scp> Risk and Disease Severity. Annals of Neurology, 2021, 89, 686-697.	2.8	10

8 Ocular Involvement Occurs Frequently at All Stages of Amyotrophic Lateral Sclerosis: Preliminary

#	Article	IF	CITATIONS
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.	1.0	24
20	Genetics of primary lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2020, 21, 28-34.	1.1	13
21	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.	1.4	35
22	PON1 is a disease modifier gene in amyotrophic lateral sclerosis: association of the Q192R polymorphism with bulbar onset and reduced survival. Neurological Sciences, 2019, 40, 1469-1473.	0.9	14
23	Psychiatric Symptoms in Amyotrophic Lateral Sclerosis: Beyond a Motor Neuron Disorder. Frontiers in Neuroscience, 2019, 13, 175.	1.4	57
24	Response to the commentary "The effect of C9orf72 intermediate repeat expansions in neurodegenerative and autoimmune diseases―by Biasiotto G and Zanella I.✰. Multiple Sclerosis and Related Disorders, 2019, 27, 79-80.	0.9	1
25	Sexuality and intimacy in ALS: systematic literature review and future perspectives. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 712-719.	0.9	10
26	Cardiovascular diseases may play a negative role in the prognosis of amyotrophic lateral sclerosis. European Journal of Neurology, 2018, 25, 861-868.	1.7	29
27	Characterization of the c9orf72 GC-rich low complexity sequence in two cohorts of Italian and Turkish ALS cases. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2018, 19, 426-431.	1.1	2
28	Elevated Global DNA Methylation Is Not Exclusive to Amyotrophic Lateral Sclerosis and Is Also Observed in Spinocerebellar Ataxia Types 1 and 2. Neurodegenerative Diseases, 2018, 18, 38-48.	0.8	27
29	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	3.8	517
30	Reconsidering the causality of TIA1 mutations in ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2018, 19, 1-3.	1.1	22
31	The Arrows and Colors Cognitive Test (ACCT): A new verbal-motor free cognitive measure for executive functions in ALS. PLoS ONE, 2018, 13, e0200953.	1.1	15
32	Cognitive-behavioral longitudinal assessment in ALS: the Italian Edinburgh Cognitive and Behavioral ALS screen (ECAS). Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2018, 19, 387-395.	1.1	34
33	ALS-associated missense and nonsense TBK1 mutations can both cause loss of kinase function. Neurobiology of Aging, 2018, 71, 266.e1-266.e10.	1.5	59
34	The Complex Interplay Between Depression/Anxiety and Executive Functioning: Insights From the ECAS in a Large ALS Population. Frontiers in Psychology, 2018, 9, 450.	1.1	14
35	The LRRK2 Variant E193K Prevents Mitochondrial Fission Upon MPP+ Treatment by Altering LRRK2 Binding to DRP1. Frontiers in Molecular Neuroscience, 2018, 11, 64.	1.4	32
36	Genotypic and Phenotypic Heterogeneity in Amyotrophic Lateral Sclerosis. , 2018, , 279-295.		3

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37	No C9orf72 repeat expansion in patients with primary progressive multiple sclerosis. Multiple Sclerosis and Related Disorders, 2018, 25, 192-195.	0.9	9
38	An eye-tracking controlled neuropsychological battery for cognitive assessment in neurological diseases. Neurological Sciences, 2017, 38, 595-603.	0.9	17
39	An eye-tracker controlled cognitive battery: overcoming verbal-motor limitations in ALS. Journal of Neurology, 2017, 264, 1136-1145.	1.8	27
40	Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. Science Translational Medicine, 2017, 9, .	5.8	129
41	Genetics of Amyotrophic Lateral Sclerosis. , 2017, , 43-59.		2
42	Use of Noninvasive Ventilation During Feeding Tube Placement. Respiratory Care, 2017, 62, 1474-1484.	0.8	14
43	Comorbidity of dementia with amyotrophic lateral sclerosis (ALS): insights from a large multicenter Italian cohort. Journal of Neurology, 2017, 264, 2224-2231.	1.8	19
44	The role of de novo mutations in the development of amyotrophic lateral sclerosis. Human Mutation, 2017, 38, 1534-1541.	1.1	13
45	A novel nonsense ATP7A pathogenic variant in a family exhibiting a variable occipital horn syndrome phenotype. Molecular Genetics and Metabolism Reports, 2017, 13, 14-17.	0.4	7
46	Factors predicting survival in ALS: a multicenter Italian study. Journal of Neurology, 2017, 264, 54-63.	1.8	96
47	Brain-Computer Interface for Clinical Purposes: Cognitive Assessment and Rehabilitation. BioMed Research International, 2017, 2017, 1-11.	0.9	83
48	Cognitive assessment in Amyotrophic Lateral Sclerosis by means of P300-Brain Computer Interface: a preliminary study. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2016, 17, 473-481.	1.1	12
49	MRI abnormalities found 1Âyear prior to symptom onset in a case of Creutzfeldt–Jakob disease. Journal of Neurology, 2016, 263, 597-599.	1.8	11
50	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1043-1048.	9.4	494
51	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	9.4	218
52	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. Nature Communications, 2016, 7, 11253.	5.8	174
53	The validation of the Italian Edinburgh Cognitive and Behavioural ALS Screen (ECAS). Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2016, 17, 489-498.	1.1	125
54	Association of a Locus in the <i>CAMTA1</i> Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. JAMA Neurology, 2016, 73, 812.	4.5	57

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55	Amyotrophic Lateral Sclerosis: Epidemiology and Risk Factors. , 2016, , 219-230.		2
56	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. Science, 2015, 347, 1436-1441.	6.0	823
57	TUBA4A gene analysis in sporadic amyotrophic lateral sclerosis: identification of novel mutations. Journal of Neurology, 2015, 262, 1376-1378.	1.8	44
58	CHCHD10mutations in Italian patients with sporadic amyotrophic lateral sclerosis: Figure 1. Brain, 2015, 138, e372-e372.	3.7	59
59	A Review of Options for Treating Sialorrhea in Amyotrophic Lateral Sclerosis. Respiratory Care, 2015, 60, 446-454.	0.8	64
60	Novel mutations support a role for Profilin 1 in the pathogenesis of ALS. Neurobiology of Aging, 2015, 36, 1602.e17-1602.e27.	1.5	87
61	Analysis of the KIFAP3 gene in amyotrophic lateral sclerosis: a multicenter survival study. Neurobiology of Aging, 2014, 35, 2420.e13-2420.e14.	1.5	16
62	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.	1.4	123
63	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. Neuron, 2014, 84, 324-331.	3.8	308
64	C9orf72 repeat expansions are restricted to the ALS-FTD spectrum. Neurobiology of Aging, 2014, 35, 936.e13-936.e17.	1.5	28
65	Amyotrophic Lateral Sclerosis: Genotypes and Phenotypes. , 2014, , 179-192.		1
66	Oligoclonal bands in the cerebrospinal fluid of amyotrophic lateral sclerosis patients with disease-associated mutations. Journal of Neurology, 2013, 260, 85-92.	1.8	24
67	Screening of the PFN1 gene in sporadic amyotrophic lateral sclerosis and in frontotemporal dementia. Neurobiology of Aging, 2013, 34, 1517.e9-1517.e10.	1.5	35
68	Analysis of hnRNPA1, A2/B1, and A3 genes in patients with amyotrophic lateral sclerosis. Neurobiology of Aging, 2013, 34, 2695.e11-2695.e12.	1.5	30
69	Randomized double-blind placebo-controlled trial of acetyl-L-carnitine for ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 397-405.	1.1	68
70	The C9ORF72 expansion mutation is a common cause of ALS+/â^'FTD in Europe and has a single founder. European Journal of Human Genetics, 2013, 21, 102-108.	1.4	201
71	<i>Ubiquilin 2</i> mutations in Italian patients with amyotrophic lateral sclerosis and frontotemporal dementia. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 183-187.	0.9	74
72	Prevalence of Huntington's disease gene CAG repeat alleles in sporadic amyotrophic lateral sclerosis patients. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 265-269.	2.3	15

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73	Mutations in the profilin 1 gene cause familial amyotrophic lateral sclerosis. Nature, 2012, 488, 499-503.	13.7	522
74	Mutational analysis of VCP gene in familial amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 630.e1-630.e2.	1.5	17
75	ATAXIN2 CAG-repeat length in Italian patients with amyotrophic lateral sclerosis: risk factor or variant phenotype? Implication for genetic testing and counseling. Neurobiology of Aging, 2012, 33, 1847.e15-1847.e21.	1.5	27
76	C9ORF72 repeat expansion in a large Italian ALS cohort: evidence of a founder effect. Neurobiology of Aging, 2012, 33, 2528.e7-2528.e14.	1.5	74
77	No association of DPP6 with amyotrophic lateral sclerosis in an Italian population. Neurobiology of Aging, 2011, 32, 966-967.	1.5	28
78	Mutational analysis of TARDBP in neurodegenerative diseases. Neurobiology of Aging, 2011, 32, 2096-2099.	1.5	43
79	VPS54 genetic analysis in ALS Italian cohort. European Journal of Neurology, 2011, 18, e41-e42.	1.7	6
80	Mutational analysis reveals the <i>FUS</i> homolog <i>TAF15</i> as a candidate gene for familial amyotrophic lateral sclerosis. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2011, 156, 285-290.	1.1	148
81	Angiogenin variants in Parkinson disease and amyotrophic lateral sclerosis. Annals of Neurology, 2011, 70, 964-973.	2.8	168
82	Novel optineurin mutations in patients with familial and sporadic amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2011, 82, 1239-1243.	0.9	86
83	RNA-binding proteins and RNA metabolism: a new scenario in the pathogenesis of Amyotrophic lateral sclerosis. Archives Italiennes De Biologie, 2011, 149, 83-99.	0.1	26
84	Genetics of familial Amyotrophic lateral sclerosis. Archives Italiennes De Biologie, 2011, 149, 65-82.	0.1	70
85	Paraoxonase gene mutations in amyotrophic lateral sclerosis. Annals of Neurology, 2010, 68, 102-107.	2.8	67
86	Protein Aggregation and Defective RNA Metabolism as Mechanisms for Motor Neuron Damage. CNS and Neurological Disorders - Drug Targets, 2010, 9, 285-296.	0.8	37
87	Stem Cells in Amyotrophic Lateral Sclerosis: Motor Neuron Protection or Replacement?. CNS and Neurological Disorders - Drug Targets, 2010, 9, 314-324.	0.8	21
88	Analysis of <i>FUS</i> gene mutation in familial amyotrophic lateral sclerosis within an Italian cohort. Neurology, 2009, 73, 1180-1185.	1.5	139
89	Reduced expression of the <i>Kinesin-Associated Protein 3</i> ( <i>KIFAP3</i> ) gene increases survival in sporadic amyotrophic lateral sclerosis. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 9004-9009.	3.3	177
90	High frequency of <i>TARDBP</i> gene mutations in Italian patients with amyotrophic lateral sclerosis. Human Mutation, 2009, 30, 688-694.	1.1	184

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91	Mutations in the <i>FUS/TLS</i> Gene on Chromosome 16 Cause Familial Amyotrophic Lateral Sclerosis. Science, 2009, 323, 1205-1208.	6.0	2,302
92	Identification of new ANG gene mutations in a large cohort of Italian patients with amyotrophic lateral sclerosis. Neurogenetics, 2008, 9, 33-40.	0.7	102
93	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. SSRN Electronic Journal, 0, , .	0.4	4
94	Parkinsonian Syndromes in Motor Neuron Disease: A Clinical Study. Frontiers in Aging Neuroscience, 0, 14, .	1.7	7
95	Expanding the phenotype of <i>TARDBP</i> mutation in a Tunisian family with clinical phenotype heterogeneity. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 0, , 1-4.	1.1	1