

# Serena Lattante

## List of Publications by Year in descending order

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61  
papers

3,064  
citations

159525

30  
h-index

168321

53  
g-index

62  
all docs

62  
docs citations

62  
times ranked

5716  
citing authors

#	ARTICLE	IF	CITATIONS
1	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , 2018, 97, 1268-1283.e6.	3.8	517
2	Loss of function of C9orf72 causes motor deficits in a zebrafish model of amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 2013, 74, 180-187.	2.8	284
3	<i>TARDBP</i> and <i>FUS</i> Mutations Associated with Amyotrophic Lateral Sclerosis: Summary and Update. <i>Human Mutation</i> , 2013, 34, 812-826.	1.1	216
4	Mutations in KANSL1 cause the 17q21.31 microdeletion syndrome phenotype. <i>Nature Genetics</i> , 2012, 44, 636-638.	9.4	148
5	P525L <i>FUS</i> mutation is consistently associated with a severe form of juvenile Amyotrophic Lateral Sclerosis. <i>Neuromuscular Disorders</i> , 2012, 22, 73-75.	0.3	124
6	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 2019, 85, 470-481.	2.8	118
7	Defining the genetic connection linking amyotrophic lateral sclerosis (ALS) with frontotemporal dementia (FTD). <i>Trends in Genetics</i> , 2015, 31, 263-273.	2.9	106
8	Contribution of major amyotrophic lateral sclerosis genes to the etiology of sporadic disease. <i>Neurology</i> , 2012, 79, 66-72.	1.5	99
9	Mutations in the 3' untranslated region of <i>FUS</i> causing <i>FUS</i> overexpression are associated with amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2013, 22, 4748-4755.	1.4	94
10	<i>ATXN2</i> trinucleotide repeat length correlates with risk of ALS. <i>Neurobiology of Aging</i> , 2017, 51, 178.e1-178.e9.	1.5	86
11	Homozygous <i>TREM2</i> mutation in a family with atypical frontotemporal dementia. <i>Neurobiology of Aging</i> , 2014, 35, 2419.e23-2419.e25.	1.5	84
12	Contribution of <i>ATXN2</i> intermediary polyQ expansions in a spectrum of neurodegenerative disorders. <i>Neurology</i> , 2014, 83, 990-995.	1.5	70
13	<i>Sqstm1</i> knock-down causes a locomotor phenotype ameliorated by rapamycin in a zebrafish model of ALS/FTLD. <i>Human Molecular Genetics</i> , 2015, 24, 1682-1690.	1.4	69
14	TRAPPC9-related autosomal recessive intellectual disability: report of a new mutation and clinical phenotype. <i>European Journal of Human Genetics</i> , 2013, 21, 229-232.	1.4	65
15	Defining the spectrum of frontotemporal dementias associated with <i>TARDBP</i> mutations. <i>Neurology: Genetics</i> , 2016, 2, e80.	0.9	56
16	The Pitt-Hopkins syndrome: Report of 16 new patients and clinical diagnostic criteria. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1536-1545.	0.7	55
17	<i>ATXN2</i> polyQ intermediate repeats are a modifier of ALS survival. <i>Neurology</i> , 2015, 84, 251-258.	1.5	52
18	hnRNPA2B1 and hnRNPA1 mutations are rare in patients with multisystem proteinopathy and frontotemporal lobar degeneration phenotypes. <i>Neurobiology of Aging</i> , 2014, 35, 934.e5-934.e6.	1.5	47

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19	Intragenic <i>KANSL1</i> mutations and chromosome 17q21.31 deletions: broadening the clinical spectrum and genotype-phenotype correlations in a large cohort of patients. <i>Journal of Medical Genetics</i> , 2015, 52, 804-814.	1.5	47
20	Rare missense variants of neuronal nicotinic acetylcholine receptor altering receptor function are associated with sporadic amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2009, 18, 3997-4006.	1.4	42
21	Primary fibroblasts cultures reveal TDP-43 abnormalities in amyotrophic lateral sclerosis patients with and without SOD1 mutations. <i>Neurobiology of Aging</i> , 2015, 36, 2005.e5-2005.e13.	1.5	42
22	LETM1 couples mitochondrial DNA metabolism and nutrient preference. <i>EMBO Molecular Medicine</i> , 2018, 10, .	3.3	41
23	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
24	Relations between C9orf72 expansion size in blood, age at onset, age at collection and transmission across generations in patients and presymptomatic carriers. <i>Neurobiology of Aging</i> , 2019, 74, 234.e1-234.e8.	1.5	38
25	TREM2 mutations are rare in a French cohort of patients with frontotemporal dementia. <i>Neurobiology of Aging</i> , 2013, 34, 2443.e1-2443.e2.	1.5	35
26	Matrin 3 variants are frequent in Italian ALS patients. <i>Neurobiology of Aging</i> , 2017, 49, 218.e1-218.e7.	1.5	35
27	ATXN1 intermediate-length polyglutamine expansions are associated with amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2018, 64, 157.e1-157.e5.	1.5	34
28	Defining the association of TMEM106B variants among frontotemporal lobar degeneration patients with GRN mutations and C9orf72 repeat expansions. <i>Neurobiology of Aging</i> , 2014, 35, 2658.e1-2658.e5.	1.5	33
29	SOD1 G93D sporadic amyotrophic lateral sclerosis (SALS) patient with rapid progression and concomitant novel ANG variant. <i>Neurobiology of Aging</i> , 2011, 32, 1924.e15-1924.e18.	1.5	32
30	Uncovering amyotrophic lateral sclerosis phenotypes: Clinical features and long-term follow-up of upper motor neuron-dominant ALS. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2011, 12, 278-282.	2.3	32
31	New ALS-Related Genes Expand the Spectrum Paradigm of Amyotrophic Lateral Sclerosis. <i>Brain Pathology</i> , 2016, 26, 266-275.	2.1	26
32	The S100A4 Transcriptional Inhibitor Niclosamide Reduces Pro-Inflammatory and Migratory Phenotypes of Microglia: Implications for Amyotrophic Lateral Sclerosis. <i>Cells</i> , 2019, 8, 1261.	1.8	24
33	Classification of familial amyotrophic lateral sclerosis by family history: effects on frequency of genes mutation. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2012, 83, 1201-1203.	0.9	22
34	Mutations in the PFN1 gene are not a common cause in patients with amyotrophic lateral sclerosis and frontotemporal lobar degeneration in France. <i>Neurobiology of Aging</i> , 2013, 34, 1709.e1-1709.e2.	1.5	21
35	Coexistence of variants in TBK1 and in other ALS-related genes elucidates an oligogenic model of pathogenesis in sporadic ALS. <i>Neurobiology of Aging</i> , 2019, 84, 239.e9-239.e14.	1.5	21
36	Syndromic Craniosynostosis Can Define New Candidate Genes for Suture Development or Result from the Non-specific Effects of Pleiotropic Genes: Rasopathies and Chromatinopathies as Examples. <i>Frontiers in Neuroscience</i> , 2017, 11, 587.	1.4	19

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37	Impairment of different protein domains causes variable clinical presentation within Pitt-Hopkins syndrome and suggests intragenic molecular syndromology of TCF4. <i>European Journal of Medical Genetics</i> , 2017, 60, 565-571.	0.7	18
38	ALS skin fibroblasts reveal oxidative stress and ERK1/2-mediated cytoplasmic localization of TDP-43. <i>Cellular Signalling</i> , 2020, 70, 109591.	1.7	18
39	High-Throughput Genetic Testing in ALS: The Challenging Path of Variant Classification Considering the ACMG Guidelines. <i>Genes</i> , 2020, 11, 1123.	1.0	15
40	Frontotemporal dementia, Parkinsonism and lower motor neuron involvement in a patient with C9ORF72 expansion. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2013, 14, 66-69.	1.1	13
41	D11Y SOD1 mutation and benign ALS: A consistent genotype-phenotype correlation. <i>Journal of the Neurological Sciences</i> , 2011, 309, 31-33.	0.3	12
42	A novel compound heterozygous <i>ALS2</i> mutation in two Italian siblings with juvenile amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2013, 14, 470-472.	1.1	12
43	A novel L67P SOD1 mutation in an Italian ALS patient. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2011, 12, 150-152.	2.3	11
44	A novel truncating variant within exon 7 of <i>KAT6B</i> associated with features of both Say-Barber-Biesecker-Young-Simpson syndrome and genitopatellar syndrome: Further evidence of a continuum in the clinical spectrum of <i>KAT6B</i> -related disorders. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 455-459.	0.7	11
45	Germline pathogenic variant in <i>PIK3CA</i> leading to symmetrical overgrowth with marked macrocephaly and mild global developmental delay. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2019, 7, e845.	0.6	11
46	Targeting S100A4 with niclosamide attenuates inflammatory and profibrotic pathways in models of amyotrophic lateral sclerosis. <i>Journal of Neuroinflammation</i> , 2021, 18, 132.	3.1	11
47	Replication of association of <i>CHRNA4</i> 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
48	Novel variants and cellular studies on patients' primary fibroblasts support a role for <i>NEK1</i> missense variants in ALS pathogenesis. <i>Human Molecular Genetics</i> , 2021, 30, 65-71.	1.4	7
49	<i>SLITRK2</i> , an X-linked modifier of the age at onset in <i>C9orf72</i> frontotemporal lobar degeneration. <i>Brain</i> , 2021, 144, 2798-2811.	3.7	7
50	Screening <i>UBQLN-2</i> in French frontotemporal lobar degeneration and frontotemporal lobar degeneration-amyotrophic lateral sclerosis patients. <i>Neurobiology of Aging</i> , 2013, 34, 2078.e5-2078.e6.	1.5	6
51	Adult phenotype in Koolen-de Vries/ <i>KANSL1</i> haploinsufficiency syndrome. <i>Journal of Medical Genetics</i> , 2022, 59, 189-195.	1.5	6
52	Wolf-Hirschhorn syndrome due to pure and translocation forms of monosomy 4p16.1. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1833-1847.	0.7	4
53	Founder effect hypothesis of D11Y SOD1 mutation in Italian amyotrophic lateral sclerosis patients. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2012, 13, 241-242.	2.3	4
54	SOD1 p.D12Y variant is associated with amyotrophic lateral sclerosis/distal myopathy spectrum. <i>European Journal of Neurology</i> , 2020, 27, 1304-1309.	1.7	4

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55	Generation and characterization of a human iPSC line from an ALS patient carrying the Q66K-MATR3 mutation. <i>Stem Cell Research</i> , 2018, 33, 146-150.	0.3	3
56	FUS mutations dominate TBK1 mutations in FUS/TBK1 double-mutant ALS/FTD pedigrees. <i>Neurogenetics</i> , 2022, 23, 59-65.	0.7	3
57	Characterization of the p.L145F and p.S135N Mutations in SOD1: Impact on the Metabolism of Fibroblasts Derived from Amyotrophic Lateral Sclerosis Patients. <i>Antioxidants</i> , 2022, 11, 815.	2.2	3
58	Peripheral neuropathy and 46XY gonadal dysgenesis: Confirmation of a heterogeneous entity. <i>Clinical Neurology and Neurosurgery</i> , 2012, 114, 748-750.	0.6	2
59	Generation of an induced pluripotent stem cell line (CSS012-A (7672)) carrying the p.G376D heterozygous mutation in the TARDBP protein. <i>Stem Cell Research</i> , 2021, 53, 102356.	0.3	1
60	Generation of an induced pluripotent stem cell line (UCSCi002-A) from a patient with a variant in TARDBP gene associated with familial amyotrophic lateral sclerosis and frontotemporal dementia. <i>Stem Cell Research</i> , 2022, 62, 102825.	0.3	1
61	Generation of an induced pluripotent stem cell line (UCSCi001-A) from a patient with early-onset amyotrophic lateral sclerosis carrying a FUS variant. <i>Stem Cell Research</i> , 2021, 55, 102461.	0.3	0