Giulia Bisogni

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/1107596/publications.pdf

Version: 2024-02-01

46 papers	1,293 citations	361413 20 h-index	377865 34 g-index

48 all docs	48 docs citations	48 times ranked	2015 citing authors

#	Article	IF	Citations
1	Antibodies to neurofascin, contactin-1, and contactin-associated protein 1 in CIDP. Neurology: Neuroimmunology and NeuroInflammation, 2020, 7, .	6.0	118
2	Contribution of major amyotrophic lateral sclerosis genes to the etiology of sporadic disease. Neurology, 2012, 79, 66-72.	1.1	99
3	Mutations in the 3â€² untranslated region of FUS causing FUS overexpression are associated with amyotrophic lateral sclerosis. Human Molecular Genetics, 2013, 22, 4748-4755.	2.9	94
4	Long-term safety and efficacy of patisiran for hereditary transthyretin-mediated amyloidosis with polyneuropathy: 12-month results of an open-label extension study. Lancet Neurology, The, 2021, 20, 49-59.	10.2	93
5	Diagnosis and Treatment of Hereditary Transthyretin Amyloidosis (hATTR) Polyneuropathy: Current Perspectives on Improving Patient Care . Therapeutics and Clinical Risk Management, 2020, Volume 16, 109-123.	2.0	78
6	Monitoring effectiveness and safety of Tafamidis in transthyretin amyloidosis in Italy: a longitudinal multicenter study in a non-endemic area. Journal of Neurology, 2016, 263, 916-924.	3.6	76
7	ATXN2 polyQ intermediate repeats are a modifier of ALS survival. Neurology, 2015, 84, 251-258.	1.1	52
8	ATTRv amyloidosis Italian Registry: clinical and epidemiological data. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 2020, 27, 259-265.	3.0	51
9	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	9.0	46
10	TTR-related amyloid neuropathy: clinical, electrophysiological and pathological findings in 15 unrelated patients. Neurological Sciences, 2013, 34, 1057-1063.	1.9	43
11	Primary fibroblasts cultures reveal TDP-43 abnormalities in amyotrophic lateral sclerosis patients with and without SOD1 mutations. Neurobiology of Aging, 2015, 36, 2005.e5-2005.e13.	3.1	42
12	Nerve ultrasound in hereditary transthyretin amyloidosis: red flags and possible progression biomarkers. Journal of Neurology, 2021, 268, 189-198.	3.6	38
13	A longitudinal study defined circulating microRNAs as reliable biomarkers for disease prognosis and progression in ALS human patients. Cell Death Discovery, 2021, 7, 4.	4.7	36
14	Matrin 3 variants are frequent in Italian ALS patients. Neurobiology of Aging, 2017, 49, 218.e1-218.e7.	3.1	35
15	ATXN1 intermediate-length polyglutamine expansions are associated with amyotrophic lateral sclerosis. Neurobiology of Aging, 2018, 64, 157.e1-157.e5.	3.1	34
16	Uncovering amyotrophic lateral sclerosis phenotypes: Clinical features and long-term follow-up of upper motor neuron-dominant ALS. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2011, 12, 278-282.	2.1	32
17	Ultrasound evaluation in transthyretinâ€related amyloid neuropathy. Muscle and Nerve, 2014, 50, 372-376.	2.2	32
18	Clinical, neurophysiological and pathological findings of HNPP patients with 17p12 deletion: A single-centre experience. Journal of the Neurological Sciences, 2014, 341, 46-50.	0.6	32

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19	Sudoscan in the evaluation and follow-up of patients and carriers with TTR mutations: experience from an Italian Centre. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 2018, 25, 242-246.	3.0	28
20	hATTR Pathology: Nerve Biopsy Results from Italian Referral Centers. Brain Sciences, 2020, 10, 780.	2.3	24
21	Recommendations for pre-symptomatic genetic testing for hereditary transthyretin amyloidosis in the era of effective therapy: a multicenter Italian consensus. Orphanet Journal of Rare Diseases, 2020, 15, 348.	2.7	22
22	Coexistence of variants in TBK1 and in other ALS-related genes elucidates an oligogenic model of pathogenesis in sporadic ALS. Neurobiology of Aging, 2019, 84, 239.e9-239.e14.	3.1	21
23	Charcot-Marie-Tooth type 2 and distal hereditary motor neuropathy: Clinical, neurophysiological and genetic findings from a single-centre experience. Clinical Neurology and Neurosurgery, 2016, 144, 67-71.	1.4	18
24	Sural nerve pathology in ALS patients: a single-centre experience. Neurological Sciences, 2012, 33, 1095-1099.	1.9	17
25	High-Throughput Genetic Testing in ALS: The Challenging Path of Variant Classification Considering the ACMG Guidelines. Genes, 2020, 11, 1123.	2.4	15
26	Progressive brachial plexus enlargement in hereditary transthyretin amyloidosis. Journal of Neurology, 2022, 269, 1905-1912.	3.6	13
27	D11Y SOD1 mutation and benign ALS: A consistent genotype-phenotype correlation. Journal of the Neurological Sciences, 2011, 309, 31-33.	0.6	12
28	Sural nerve biopsy in peripheral neuropathies: 30-year experience from a single center. Neurological Sciences, 2020, 41, 341-346.	1.9	12
29	Restless Leg Syndrome in Different Types of Demyelinating Neuropathies: A Single-Center Pilot Study. Journal of Clinical Sleep Medicine, 2013, 09, 945-949.	2.6	12
30	Pathological Findings in Chronic Inflammatory Demyelinating Polyradiculoneuropathy: A Single-Center Experience. Brain Sciences, 2020, 10, 383.	2.3	10
31	Gastrointestinal Manifestations in Hereditary Transthyretin Amyloidosis: a Single-Centre Experience. Journal of Gastrointestinal and Liver Diseases, 2020, 29, 339-343.	0.9	10
32	Patisiran in hATTR Amyloidosis: Six-Month Latency Period before Efficacy. Brain Sciences, 2021, 11, 515.	2.3	8
33	Psychosocial burden and professional and social support in patients with hereditary transthyretin amyloidosis (ATTRv) and their relatives in Italy. Orphanet Journal of Rare Diseases, 2021, 16, 163.	2.7	8
34	Novel variants and cellular studies on patientsâ€™ primary fibroblasts support a role for NEK1 missense variants in ALS pathogenesis. Human Molecular Genetics, 2021, 30, 65-71.	2.9	7
35	Clinical, electrophysiological and pathological findings in a patient with Charcotâ€“Marieâ€“Tooth disease 4D caused by the NDRG1 Lom mutation. Journal of the Neurological Sciences, 2014, 345, 271-273.	0.6	6
36	Reassessing IVIg therapy in chronic inflammatory demyelinating polyradiculoneuropathy during COVID-19: a chance to verify the need for chronic maintenance therapy. Neurological Sciences, 2021, 42, 787-789.	1.9	6

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37	SOD1 p.D12Y variant is associated with amyotrophic lateral sclerosis/distal myopathy spectrum. European Journal of Neurology, 2020, 27, 1304-1309.	3.3	4
38	Distinct lymphocytes subsets in IgM-related neuropathy: clinical-immunological correlations. Neurological Sciences, 2015, 36, 303-308.	1.9	2
39	‘White Nails'. European Neurology, 2015, 73, 89-89.	1.4	2
40	Small Fibre Involvement in Multifocal Motor Neuropathy Explored with Sudoscan: A Single-Centre Experience. Diagnostics, 2020, 10, 755.	2.6	2
41	Progressive axonal polyneuropathy in a mitochondrial disorder: an uncommon association with familial amyloid neuropathy. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 2018, 25, 261-262.	3.0	1
42	Isolated light chain deposition disease neuropathy in a patient with multiple myeloma. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 2020, 27, 67-68.	3.0	1
43	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. Stem Cell Research, 2022, 62, 102825.	0.7	1
44	Skin Changes in POEMS Syndrome. European Neurology, 2015, 73, 112-112.	1.4	0
45	Thr124Met myelin protein zero mutation mimicking motor neuron disease. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2021, , 1-6.	1.7	0
46	Generation of an induced pluripotent stem cell line (UCSCi001-A) from a patient with early-onset amyotrophic lateral sclerosis carrying a FUS variant. Stem Cell Research, 2021, 55, 102461.	0.7	0