Andrea Cortese

List of Publications by Year
in descending order

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100 papers	3,688 citations	172457 29 h-index	149698 56 g-index
107 all docs	107 docs citations	107 times ranked	6034 citing authors

#	Article	IF	Citations
1	Repurposing Diflunisal for Familial Amyloid Polyneuropathy. JAMA - Journal of the American Medical Association, 2013, 310, 2658.	7.4	551
2	Biallelic expansion of an intronic repeat in RFC1 is a common cause of late-onset ataxia. Nature Genetics, 2019, 51, 649-658.	21.4	338
3	Doxycycline plus tauroursodeoxycholic acid for transthyretin amyloidosis: a phase II study. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 2012, 19, 34-36.	3.0	184
4	Autoantibodies to nodal isoforms of neurofascin in chronic inflammatory demyelinating polyneuropathy. Brain, 2017, 140, 1851-1858.	7.6	167
5	Cerebellar ataxia, neuropathy, vestibular areflexia syndrome due to RFC1 repeat expansion. Brain, 2020, 143, 480-490.	7.6	140
6	Antibodies to neurofascin, contactin-1, and contactin-associated protein 1 in CIDP. Neurology: Neuroimmunology and NeuroInflammation, 2020, 7, .	6.0	118
7	Atypical CIDP: diagnostic criteria, progression and treatment response. Data from the Italian CIDP Database. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 125-132.	1.9	108
8	Biallelic mutations in SORD cause a common and potentially treatable hereditary neuropathy with implications for diabetes. Nature Genetics, 2020, 52, 473-481.	21.4	97
9	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
10	Natural History, Phenotypic Spectrum, and Discriminative Features of Multisystemic RFC1 Disease. Neurology, 2021, 96, e1369-e1382.	1.1	93
11	Diagnostic challenges in hereditary transthyretin amyloidosis with polyneuropathy: avoiding misdiagnosis of a treatable hereditary neuropathy. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 457-458.	1.9	83
12	Correlation of clinical and molecular features in spinal bulbar muscular atrophy. Neurology, 2014, 82, 2077-2084.	1.1	76
13	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
14	Targeted next-generation sequencing panels in the diagnosis of Charcot-Marie-Tooth disease. Neurology, 2020, 94, e51-e61.	1.1	71
15	Comprehensive genetic diagnosis of tandem repeat expansion disorders with programmable targeted nanopore sequencing. Science Advances, 2022, 8, eabm5386.	10.3	68
16	Longitudinal observational study of sporadic inclusion body myositis: Implications for clinical trials. Neuromuscular Disorders, 2013, 23, 404-412.	0.6	63
17	RFC1 expansions are a common cause of idiopathic sensory neuropathy. Brain, 2021, 144, 1542-1550.	7.6	63
18	Combined central and peripheral demyelination: Clinical features, diagnostic findings, and treatment. Journal of the Neurological Sciences, 2016, 363, 182-187.	0.6	55

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19	A novel RFC1 repeat motif (ACAGG) in two Asia-Pacific CANVAS families. Brain, 2020, 143, 2904-2910.	7.6	53
20	Anti-NF155 chronic inflammatory demyelinating polyradiculoneuropathy strongly associates to HLA-DRB15. Journal of Neuroinflammation, 2017, 14, 224.	7.2	50
21	A MÄori specific RFC1 pathogenic repeat configuration in CANVAS, likely due to a founder allele. Brain, 2020, 143, 2673-2680.	7.6	45
22	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. Brain, 2019, 142, 2948-2964.	7.6	43
23	Assessing mNIS+7_{Ionis} and international neurologists' proficiency in a familial amyloidotic polyneuropathy trial. Muscle and Nerve, 2017, 56, 901-911.	2.2	42
24	Diagnosis and therapy of acute disseminated encephalomyelitis and its variants. Expert Review of Neurotherapeutics, 2016, 16, 83-101.	2.8	40
25	Truncating Mutations in UBAP1 Cause Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2019, 104, 767-773.	6.2	39
26	Novel CLN3 mutation causing autophagic vacuolar myopathy. Neurology, 2014, 82, 2072-2076.	1.1	37
27	Widespread RNA metabolism impairment in sporadic inclusion body myositis TDP43-proteinopathy. Neurobiology of Aging, 2014, 35, 1491-1498.	3.1	36
28	Botulinum Toxin Is Effective in the Management of Neurogenic Dysphagia. Clinical-Electrophysiological Findings and Tips on Safety in Different Neurological Disorders. Frontiers in Pharmacology, 2017, 8, 80.	3.5	36
29	Air pollution is associated to the multiple sclerosis inflammatory activity as measured by brain MRI. Multiple Sclerosis Journal, 2018, 24, 1578-1584.	3.0	35
30	Neurofascin-155 as a putative antigen in combined central and peripheral demyelination. Neurology: Neuroimmunology and NeuroInflammation, 2016, 3, e238.	6.0	32
31	RFC1 repeat expansion in Japanese patients with late-onset cerebellar ataxia. Journal of Human Genetics, 2020, 65, 1143-1147.	2.3	32
32	Motor neuron pathology in CANVAS due to RFC1 expansions. Brain, 2022, 145, 2121-2132.	7.6	32
33	Plasma neurofilament light chain: an early biomarker for hereditary ATTR amyloid polyneuropathy. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 2020, 27, 97-102.	3.0	31
34	Clinical and Laboratory Features in Anti-NF155 Autoimmune Nodopathy. Neurology: Neuroimmunology and NeuroInflammation, 2022, 9, .	6.0	30
35	Air pollution as a contributor to the inflammatory activity of multiple sclerosis. Journal of Neuroinflammation, 2020, 17, 334.	7.2	28
36	Risk factors for chronic inflammatory demyelinating polyradiculoneuropathy (CIDP): antecedent events, lifestyle and dietary habits. Data from the Italian CIDP Database. European Journal of Neurology, 2020, 27, 136-143.	3.3	27

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37	<scp>RFC1</scp> Intronic Repeat Expansions Absent in Pathologically Confirmed Multiple Systems Atrophy. Movement Disorders, 2020, 35, 1277-1279.	3.9	26
38	RFC1 expansions can mimic hereditary sensory neuropathy with cough and SjÃ¶gren syndrome. Brain, 2020, 143, e82-e82.	7.6	25
39	RFC1-related ataxia is a mimic of early multiple system atrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 444-446.	1.9	25
40	Altered <scp>TDP</scp>â€43â€dependent splicing in <scp>HSPB</scp>8â€related distal hereditary motor neuropathy and myofibrillar myopathy. European Journal of Neurology, 2018, 25, 154-163.	3.3	24
41	Novel mutations in human and mouse SCN4A implicate AMPK in myotonia and periodic paralysis. Brain, 2014, 137, 3171-3185.	7.6	23
42	Frequency of diabetes and other comorbidities in chronic inflammatory demyelinating polyradiculoneuropathy and their impact on clinical presentation and response to therapy. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 1092-1099.	1.9	22
43	Copper deficiency in Wilson's disease: Peripheral neuropathy and myelodysplastic syndrome complicating zinc treatment. Movement Disorders, 2011, 26, 1361-1362.	3.9	21
44	Cell environment shapes TDP-43 function with implications in neuronal and muscle disease. Communications Biology, 2022, 5, 314.	4.4	21
45	An ancestral 10-bp repeat expansion in VWA1 causes recessive hereditary motor neuropathy. Brain, 2021, 144, 584-600.	7.6	20
46	CANVAS: a late onset ataxia due to biallelic intronic AAGGG expansions. Journal of Neurology, 2021, 268, 1119-1126.	3.6	19
47	Theta-Burst Stimulation of the Cerebellum Interferes with Internal Representations of Sensory-Motor Information Related to Eye Movements in Humans. Cerebellum, 2011, 10, 711-719.	2.5	17
48	Relevance of diagnostic investigations in chronic inflammatory demyelinating poliradiculoneuropathy: Data from the Italian CIDP database. Journal of the Peripheral Nervous System, 2020, 25, 152-161.	3.1	15
49	Chronic inflammatory demyelinating polyradiculoneuropathy: can a diagnosis be made in patients not fulfilling electrodiagnostic criteria?. European Journal of Neurology, 2021, 28, 620-629.	3.3	15
50	Multiple Coronary Arteryâ€“Left Ventricle Microfistulae and Spongy Myocardium. Circulation, 2007, 116, e81-4.	1.6	14
51	Cerebellar ataxia, neuropathy and vestibular areflexia syndrome (CANVAS): genetic and clinical aspects. Practical Neurology, 2022, 22, 14-18.	1.1	14
52	RFC1 repeat expansions: A recurrent cause of sensory and autonomic neuropathy with cough and ataxia. European Journal of Neurology, 2022, 29, 2156-2161.	3.3	14
53	Guillainâ€“BarrÃ© syndrome associated with the D222E variant of the 2009 pandemic influenza A (H1N1) virus: Case report and review of the literature. Journal of the Neurological Sciences, 2012, 312, 173-176.	0.6	13
54	IGHMBP2 mutation associated with organ-specific autonomic dysfunction. Neuromuscular Disorders, 2018, 28, 1012-1015.	0.6	13

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55	Varicella zoster virusâ€“associated polyradiculoneuritis. Neurology, 2009, 73, 1334-1335.	1.1	12
56	Predictors of outcome in a large retrospective cohort of patients with transverse myelitis. Multiple Sclerosis Journal, 2018, 24, 1743-1752.	3.0	12
57	Autosomal dominant optic atrophy and cataract â€œplusâ€•phenotype including axonal neuropathy. Neurology: Genetics, 2019, 5, e322.	1.9	12
58	Mutation in RNF170 causes sensory ataxic neuropathy with vestibular areflexia: a CANVAS mimic. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 1237-1238.	1.9	11
59	Liver cirrhosis and rhino-orbital mucormycosis, a possible but rare association: description of a clinical case and literature review. Brazilian Journal of Infectious Diseases, 2009, 13, 314-6.	0.6	10
60	Graphical modelling of molecular networks underlying sporadic inclusion body myositis. Molecular BioSystems, 2013, 9, 1736.	2.9	10
61	A CADM3 variant causes Charcot-Marie-Tooth disease with marked upper limb involvement. Brain, 2021, 144, 1197-1213.	7.6	10
62	Biliodigestive anastomosis with circular mechanical device after pancreatoduodenectomy: our experience. Updates in Surgery, 2011, 63, 253-257.	2.0	9
63	Diagnostics of anti-MAG antibody polyneuropathy. Neurological Sciences, 2017, 38, 249-252.	1.9	9
64	Charcot-Marie-Tooth disease type 2CC due toNEFHvariants causes a progressive, non-length-dependent, motor-predominant phenotype. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, 48-56.	1.9	9
65	Frequency and clinical correlates of anti-nerve antibodies in a large population of CIDP patients included in the Italian database. Neurological Sciences, 2022, 43, 3939-3947.	1.9	9
66	Diagnostics of dysimmune peripheral neuropathies. Neurological Sciences, 2017, 38, 243-247.	1.9	8
67	Remarkable Rituximab Response on Tremor Related to Acuteâ€Onset Chronic Inflammatory Demyelinating Polyradiculoneuropathy in an Antineurofascin155 Immunoglobulin G4â€“Seropositive Patient. Movement Disorders Clinical Practice, 2018, 5, 559-560.	1.5	8
68	Genetic and phenotypic characterisation of inherited myopathies in a tertiary neuromuscular centre. Neuromuscular Disorders, 2019, 29, 747-757.	0.6	8
69	MR microneurography and quantitative T2 and DP measurements of the distal tibial nerve in CIDP. Journal of the Neurological Sciences, 2019, 400, 15-20.	0.6	8
70	Investigating RFC1 expansions in sporadic amyotrophic lateral sclerosis. Journal of the Neurological Sciences, 2021, 430, 118061.	0.6	8
71	Contactin-1 Antibodies Link Autoimmune Neuropathies to Nephrotic Syndrome. SSRN Electronic Journal, 0, , .	0.4	8
72	Multiple memory-guided saccades: movement memory improves the accuracy of memory-guided saccades. Progress in Brain Research, 2008, 171, 425-427.	1.4	6

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73	Bosentan and Sildenafil in the Treatment of HIV-Associated Pulmonary Hypertension. Gastroenterology Insights, 2011, 3, e14.	1.2	6
74	Characterization of three novel pathogenic SLC40A1 mutations and genotype/phenotype correlations in 7 Italian families with type 4 hereditary hemochromatosis. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 464-470.	3.8	6
75	Long read sequencing overcomes challenges in the diagnosis of <scp>SORD</scp> neuropathy. Journal of the Peripheral Nervous System, 2022, 27, 120-126.	3.1	6
76	Efficacy of rituximab as third-line therapy in combined central and peripheral demyelination. Neurology: Clinical Practice, 2017, 7, 534-537.	1.6	5
77	Impact of environmental factors and physical activity on disability and quality of life in CIDP. Journal of Neurology, 2020, 267, 2683-2691.	3.6	4
78	Unusual upper limb features in SORD neuropathy. Journal of the Peripheral Nervous System, 2022, 27, 175-177.	3.1	4
79	Laryngeal and phrenic nerve involvement in a patient with hereditary neuropathy with liability to pressure palsies (HNPP). Neuromuscular Disorders, 2016, 26, 455-458.	0.6	3
80	The neurophysiological lesson from the Italian CIDP database. Neurological Sciences, 2021, , 1.	1.9	3
81	Late onset oculopharyngeal muscular dystrophy with prominent neurogenic features and short GCG trinucleotide expansion. Muscle and Nerve, 2011, 43, 141-142.	2.2	2
82	Severe cognitive impairment in a patient with CMT2A. Journal of the Peripheral Nervous System, 2018, 23, 147-148.	3.1	2
83	Absence of pathogenic mutations in CD59 in chronic inflammatory demyelinating polyradiculoneuropathy. PLoS ONE, 2019, 14, e0212647.	2.5	2
84	Reply to: â€œDopaâ€Responsive Parkinsonism in a Patient With Homozygous RFC1 Expansionsâ€. Movement Disorders, 2020, 35, 1890-1891.	3.9	2
85	Impact of multiple sclerosis risk loci in postinfectious neurological syndromes. Multiple Sclerosis and Related Disorders, 2020, 44, 102326.	2.0	2
86	Monitoring late complications of zinc treatment in Wilson's disease. Reply to the letter: Copper deficiency in Wilson's disease: An avoidable complication of treatment. Movement Disorders, 2011, 26, 2449-2450.	3.9	1
87	Functional validation of non-coding variants of GJB1 in patients with CMTX1. Neuromuscular Disorders, 2017, 27, S25.	0.6	1
88	Variable echo time imaging for detecting the short T2* components of the sciatic nerve: a validation study. Magnetic Resonance Materials in Physics, Biology, and Medicine, 2021, 34, 411-419.	2.0	1
89	Bedside Head Impulse Test: A Useful Tool for Patients With Sensory Ataxia. Neurology: Genetics, 2021, 7, e541.	1.9	1
90	Diagnosis of Anorectal Disease. , 2010, , 91-97.		0

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91	1130â€...Clinical features and clinical course of sporadic inclusion body myositis (IBM): a prospective cohort study: IBM-net. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, e1.101-e1.	1.9	0
92	G.P.191. Neuromuscular Disorders, 2014, 24, 867.	0.6	0
93	Reply. Annals of Thoracic Surgery, 2016, 101, 826-827.	1.3	0
94	Clinical features and genetic findings in patients with Charcot Marie Tooth Disease Type 2 (CMT2) due to LRSAM1 mutation. Neuromuscular Disorders, 2017, 27, S25.	0.6	0
95	Frequency of genetic variants in Charcot-Marie-Tooth disease: how many is too many?. Neuromuscular Disorders, 2018, 28, S22.	0.6	0
96	Cement Emboli and Subsequent Traumatic Hemothorax as a Rare Complication of Vertebroplasty. , 2019, , .		0
97	Towards a standardised analysis of CSF in inflammatory neuropathies. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 916-916.	1.9	0
98	An uncommon case of recumbent dyspnea - Have a look to the diaphragm. , 2016, , .		0
99	Biallelic mutations in sord are a common cause of potentially treatable genetic neuropathy. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, A14.2-A14.	1.9	0
100	Charcot-Marie-Tooth disease secondary to biallelic mutations in SORD. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, A96.1-A96.	1.9	0