Andrea Cortese

List of Publications by Year
in descending order

100	3,688	172386	149623
papers	citations	29 h-index	56 g-index

107	107	107	6034
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Cerebellar ataxia, neuropathy and vestibular areflexia syndrome (CANVAS): genetic and clinical aspects. Practical Neurology, 2022, 22, 14-18.	0.5	14
2	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.	0.9	9
3	Clinical and Laboratory Features in Anti-NF155 Autoimmune Nodopathy. Neurology: Neuroimmunology and NeuroInflammation, 2022, 9, .	3.1	30
4	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.	0.9	9
5	Long read sequencing overcomes challenges in the diagnosis of <scp>SORD</scp> neuropathy. Journal of the Peripheral Nervous System, 2022, 27, 120-126.	1.4	6
6	Comprehensive genetic diagnosis of tandem repeat expansion disorders with programmable targeted nanopore sequencing. Science Advances, 2022, 8, eabm5386.	4.7	68
7	RFC1 repeat expansions: A recurrent cause of sensory and autonomic neuropathy with cough and ataxia. European Journal of Neurology, 2022, 29, 2156-2161.	1.7	14
8	Cell environment shapes TDP-43 function with implications in neuronal and muscle disease. Communications Biology, 2022, 5, 314.	2.0	21
9	Motor neuron pathology in CANVAS due to RFC1 expansions. Brain, 2022, 145, 2121-2132.	3.7	32
10	Unusual upper limb features in SORD neuropathy. Journal of the Peripheral Nervous System, 2022, 27, 175-177.	1.4	4
11	Biallelic mutations in sord are a common cause of potentially treatable genetic neuropathy. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, A14.2-A14.	0.9	0
12	Charcot-Marie-Tooth disease secondary to biallelic mutations in SORD. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, A96.1-A96.	0.9	0
13	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.	4.9	93
14	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.	1.1	1
15	Chronic inflammatory demyelinating polyradiculoneuropathy: can a diagnosis be made in patients not fulfilling electrodiagnostic criteria?. European Journal of Neurology, 2021, 28, 620-629.	1.7	15
16	CANVAS: a late onset ataxia due to biallelic intronic AAGGG expansions. Journal of Neurology, 2021, 268, 1119-1126.	1.8	19
17	Natural History, Phenotypic Spectrum, and Discriminative Features of Multisystemic RFC1 Disease. Neurology, 2021, 96, e1369-e1382.	1.5	93
18	An ancestral 10-bp repeat expansion in VWA1 causes recessive hereditary motor neuropathy. Brain, 2021, 144, 584-600.	3.7	20

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19	RFC1-related ataxia is a mimic of early multiple system atrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 444-446.	0.9	25
20	A CADM3 variant causes Charcot-Marie-Tooth disease with marked upper limb involvement. Brain, 2021, 144, 1197-1213.	3.7	10
21	The neurophysiological lesson from the Italian CIDP database. Neurological Sciences, 2021, , 1.	0.9	3
22	Towards a standardised analysis of CSF in inflammatory neuropathies. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 916-916.	0.9	0
23	RFC1 expansions are a common cause of idiopathic sensory neuropathy. Brain, 2021, 144, 1542-1550.	3.7	63
24	Investigating RFC1 expansions in sporadic amyotrophic lateral sclerosis. Journal of the Neurological Sciences, 2021, 430, 118061.	0.3	8
25	Bedside Head Impulse Test: A Useful Tool for Patients With Sensory Ataxia. Neurology: Genetics, 2021, 7, e541.	0.9	1
26	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.	1.7	27
27	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.	1.4	31
28	RFC1 repeat expansion in Japanese patients with late-onset cerebellar ataxia. Journal of Human Genetics, 2020, 65, 1143-1147.	1.1	32
29	A novel RFC1 repeat motif (ACAGG) in two Asia-Pacific CANVAS families. Brain, 2020, 143, 2904-2910.	3.7	53
30	Reply to: â€œDopaâ€Responsive Parkinsonism in a Patient With Homozygous RFC1 Expansionsâ€. Movement Disorders, 2020, 35, 1890-1891.	2.2	2
31	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.	0.9	22
32	RFC1 expansions can mimic hereditary sensory neuropathy with cough and SjÃ¶gren syndrome. Brain, 2020, 143, e82-e82.	3.7	25
33	Mutation in RNF170 causes sensory ataxic neuropathy with vestibular areflexia: a CANVAS mimic. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 1237-1238.	0.9	11
34	A MÄori specific RFC1 pathogenic repeat configuration in CANVAS, likely due to a founder allele. Brain, 2020, 143, 2673-2680.	3.7	45
35	Air pollution as a contributor to the inflammatory activity of multiple sclerosis. Journal of Neuroinflammation, 2020, 17, 334.	3.1	28
36	Biallelic mutations in SORD cause a common and potentially treatable hereditary neuropathy with implications for diabetes. Nature Genetics, 2020, 52, 473-481.	9.4	97

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37	Impact of environmental factors and physical activity on disability and quality of life in CIDP. Journal of Neurology, 2020, 267, 2683-2691.	1.8	4
38	Impact of multiple sclerosis risk loci in postinfectious neurological syndromes. Multiple Sclerosis and Related Disorders, 2020, 44, 102326.	0.9	2
39	Antibodies to neurofascin, contactin-1, and contactin-associated protein 1 in CIDP. Neurology: Neuroimmunology and NeuroInflammation, 2020, 7, .	3.1	118
40	Cerebellar ataxia, neuropathy, vestibular areflexia syndrome due to RFC1 repeat expansion. Brain, 2020, 143, 480-490.	3.7	140
41	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.	1.4	15
42	<scp>RFC1</scp> Intronic Repeat Expansions Absent in Pathologically Confirmed Multiple Systems Atrophy. Movement Disorders, 2020, 35, 1277-1279.	2.2	26
43	Targeted next-generation sequencing panels in the diagnosis of Charcot-Marie-Tooth disease. Neurology, 2020, 94, e51-e61.	1.5	71
44	Genetic and phenotypic characterisation of inherited myopathies in a tertiary neuromuscular centre. Neuromuscular Disorders, 2019, 29, 747-757.	0.3	8
45	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. Brain, 2019, 142, 2948-2964.	3.7	43
46	Autosomal dominant optic atrophy and cataract â€œplusâ€•phenotype including axonal neuropathy. Neurology: Genetics, 2019, 5, e322.	0.9	12
47	MR microneurography and quantitative T2 and DP measurements of the distal tibial nerve in CIDP. Journal of the Neurological Sciences, 2019, 400, 15-20.	0.3	8
48	Biallelic expansion of an intronic repeat in RFC1 is a common cause of late-onset ataxia. Nature Genetics, 2019, 51, 649-658.	9.4	338
49	Truncating Mutations in UBAP1 Cause Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2019, 104, 767-773.	2.6	39
50	Absence of pathogenic mutations in CD59 in chronic inflammatory demyelinating polyradiculoneuropathy. PLoS ONE, 2019, 14, e0212647.	1.1	2
51	Cement Emboli and Subsequent Traumatic Hemothorax as a Rare Complication of Vertebroplasty. , 2019, , .		0
52	Atypical CIDP: diagnostic criteria, progression and treatment response. Data from the Italian CIDP Database. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 125-132.	0.9	108
53	Severe cognitive impairment in a patient with CMT2A. Journal of the Peripheral Nervous System, 2018, 23, 147-148.	1.4	2
54	Predictors of outcome in a large retrospective cohort of patients with transverse myelitis. Multiple Sclerosis Journal, 2018, 24, 1743-1752.	1.4	12

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55	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.	1.7	24
56	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.	1.8	6
57	Air pollution is associated to the multiple sclerosis inflammatory activity as measured by brain MRI. Multiple Sclerosis Journal, 2018, 24, 1578-1584.	1.4	35
58	Frequency of genetic variants in Charcot-Marie-Tooth disease: how many is too many?. Neuromuscular Disorders, 2018, 28, S22.	0.3	0
59	IGHMBP2 mutation associated with organ-specific autonomic dysfunction. Neuromuscular Disorders, 2018, 28, 1012-1015.	0.3	13
60	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.	0.8	8
61	Assessing mNIS+7_{Ionis} and international neurologists' proficiency in a familial amyloidotic polyneuropathy trial. Muscle and Nerve, 2017, 56, 901-911.	1.0	42
62	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.	0.9	83
63	Functional validation of non-coding variants of GJB1 in patients with CMTX1. Neuromuscular Disorders, 2017, 27, S25.	0.3	1
64	Autoantibodies to nodal isoforms of neurofascin in chronic inflammatory demyelinating polyneuropathy. Brain, 2017, 140, 1851-1858.	3.7	167
65	Diagnostics of anti-MAG antibody polyneuropathy. Neurological Sciences, 2017, 38, 249-252.	0.9	9
66	Diagnostics of dysimmune peripheral neuropathies. Neurological Sciences, 2017, 38, 243-247.	0.9	8
67	Clinical features and genetic findings in patients with Charcot Marie Tooth Disease Type 2 (CMT2) due to LRSAM1 mutation. Neuromuscular Disorders, 2017, 27, S25.	0.3	0
68	Efficacy of rituximab as third-line therapy in combined central and peripheral demyelination. Neurology: Clinical Practice, 2017, 7, 534-537.	0.8	5
69	Anti-NF155 chronic inflammatory demyelinating polyradiculoneuropathy strongly associates to HLA-DRB15. Journal of Neuroinflammation, 2017, 14, 224.	3.1	50
70	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.	1.6	36
71	Laryngeal and phrenic nerve involvement in a patient with hereditary neuropathy with liability to pressure palsies (HNPP). Neuromuscular Disorders, 2016, 26, 455-458.	0.3	3
72	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.	1.8	76

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73	Neurofascin-155 as a putative antigen in combined central and peripheral demyelination. Neurology: Neuroimmunology and NeuroInflammation, 2016, 3, e238.	3.1	32
74	Reply. Annals of Thoracic Surgery, 2016, 101, 826-827.	0.7	0
75	Combined central and peripheral demyelination: Clinical features, diagnostic findings, and treatment. Journal of the Neurological Sciences, 2016, 363, 182-187.	0.3	55
76	Diagnosis and therapy of acute disseminated encephalomyelitis and its variants. Expert Review of Neurotherapeutics, 2016, 16, 83-101.	1.4	40
77	An uncommon case of recumbent dyspnea - Have a look to the diaphragm. , 2016, , .		0
78	Novel mutations in human and mouse SCN4A implicate AMPK in myotonia and periodic paralysis. Brain, 2014, 137, 3171-3185.	3.7	23
79	Correlation of clinical and molecular features in spinal bulbar muscular atrophy. Neurology, 2014, 82, 2077-2084.	1.5	76
80	G.P.191. Neuromuscular Disorders, 2014, 24, 867.	0.3	0
81	Widespread RNA metabolism impairment in sporadic inclusion body myositis TDP43-proteinopathy. Neurobiology of Aging, 2014, 35, 1491-1498.	1.5	36
82	Novel CLN3 mutation causing autophagic vacuolar myopathy. Neurology, 2014, 82, 2072-2076.	1.5	37
83	Longitudinal observational study of sporadic inclusion body myositis: Implications for clinical trials. Neuromuscular Disorders, 2013, 23, 404-412.	0.3	63
84	Graphical modelling of molecular networks underlying sporadic inclusion body myositis. Molecular BioSystems, 2013, 9, 1736.	2.9	10
85	Repurposing Diflunisal for Familial Amyloid Polyneuropathy. JAMA - Journal of the American Medical Association, 2013, 310, 2658.	3.8	551
86	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.	0.9	0
87	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.	1.4	184
88	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.	0.3	13
89	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.	1.4	17
90	Biliodigestive anastomosis with circular mechanical device after pancreatoduodenectomy: our experience. Updates in Surgery, 2011, 63, 253-257.	0.9	9

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91	Copper deficiency in Wilson's disease: Peripheral neuropathy and myelodysplastic syndrome complicating zinc treatment. Movement Disorders, 2011, 26, 1361-1362.	2.2	21
92	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.	2.2	1
93	Late onset oculopharyngeal muscular dystrophy with prominent neurogenic features and short GCG trinucleotide expansion. Muscle and Nerve, 2011, 43, 141-142.	1.0	2
94	Bosentan and Sildenafil in the Treatment of HIV-Associated Pulmonary Hypertension. Gastroenterology Insights, 2011, 3, e14.	0.7	6
95	Diagnosis of Anorectal Disease. , 2010, , 91-97.		0
96	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.	0.3	10
97	Varicella zoster virusâ€“associated polyradiculoneuritis. Neurology, 2009, 73, 1334-1335.	1.5	12
98	Multiple memory-guided saccades: movement memory improves the accuracy of memory-guided saccades. Progress in Brain Research, 2008, 171, 425-427.	0.9	6
99	Multiple Coronary Arteryâ€“Left Ventricle Microfistulae and Spongy Myocardium. Circulation, 2007, 116, e81-4.	1.6	14
100	Contactin-1 Antibodies Link Autoimmune Neuropathies to Nephrotic Syndrome. SSRN Electronic Journal, 0, , .	0.4	8