Alessandro Filla

List of Publications by Year
in descending order

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51 papers	2,272 citations	257450 24 h-index	223800 46 g-index
53 all docs	53 docs citations	53 times ranked	2362 citing authors

#	Article	IF	Citations
1	Episodic ataxia and severe infantile phenotype in spinocerebellar ataxia type 14: expansion of the phenotype and novel mutations. Journal of Neurology, 2022, 269, 1476-1484.	3.6	7
2	Levels of Neurofilament Light at the Preataxic and Ataxic Stages of Spinocerebellar Ataxia Type 1. Neurology, 2022, 98, .	1.1	15
3	Screening for RFC-1 pathological expansion in late-onset ataxias: a contribution to the differential diagnosis. Journal of Neurology, 2022, 269, 5431-5435.	3.6	4
4	Natural History, Phenotypic Spectrum, and Discriminative Features of Multisystemic RFC1 Disease. Neurology, 2021, 96, e1369-e1382.	1.1	93
5	Progressive myoclonus epilepsiesâ€"Residual unsolved cases have marked genetic heterogeneity including dolichol-dependent protein glycosylation pathway genes. American Journal of Human Genetics, 2021, 108, 722-738.	6.2	41
6	NGS in Hereditary Ataxia: When Rare Becomes Frequent. International Journal of Molecular Sciences, 2021, 22, 8490.	4.1	12
7	Variants in ATP6V0A1 cause progressive myoclonus epilepsy and developmental and epileptic encephalopathy. Brain Communications, 2021, 3, fcab245.	3.3	10
8	Conversion of individuals at risk for spinocerebellar ataxia types 1, 2, 3, and 6 to manifest ataxia (RISCA): a longitudinal cohort study. Lancet Neurology, The, 2020, 19, 738-747.	10.2	41
9	New AARS2 Mutations in Two Siblings With Tremor, Downbeat Nystagmus, and Primary Amenorrhea: A Benign Phenotype Without Leukoencephalopathy. Movement Disorders Clinical Practice, 2020, 7, 684-687.	1.5	8
10	Ataxia-myoclonus syndrome due to a novel homozygous ATP13A2 mutation. Parkinsonism and Related Disorders, 2020, 76, 42-43.	2.2	7
11	Neurofilaments in spinocerebellar ataxia type 3: blood biomarkers at the preataxic and ataxic stage in humans and mice. EMBO Molecular Medicine, 2020, 12, e11803.	6.9	73
12	Biâ€allelic mutations in HARS1 severely impair histidylâ€tRNA synthetase expression and enzymatic activity causing a novel multisystem ataxic syndrome. Human Mutation, 2020, 41, 1232-1237.	2.5	15
13	Spinocerebellar ataxia type 48: last but not least. Neurological Sciences, 2020, 41, 2423-2432.	1.9	31
14	Overt Hypogonadism May Not Be a Sentinel Sign of RING Finger Protein 216: Two Novel Mutations Associated with Ataxia, Chorea, and Fertility. Movement Disorders Clinical Practice, 2019, 6, 724-726.	1.5	12
15	Prediction of Survival With Longâ€Term Disease Progression in Most Common Spinocerebellar Ataxia. Movement Disorders, 2019, 34, 1220-1227.	3.9	14
16	Spinocerebellar ataxia type 2â€"neuronopathy or neuropathy?. Muscle and Nerve, 2019, 60, 271-278.	2.2	14
17	Longitudinal study of a cohort of MSA-C patients in South Italy: survival and clinical features. Neurological Sciences, 2019, 40, 2105-2109.	1.9	11
18	Degenerative and acquired sporadic adult onset ataxia. Neurological Sciences, 2019, 40, 1335-1342.	1.9	26

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19	Survival in patients with spinocerebellar ataxia types 1, 2, 3, and 6 (EUROSCA): a longitudinal cohort study. Lancet Neurology, The, 2018, 17, 327-334.	10.2	69
20	Long-term evolution of patient-reported outcome measures in spinocerebellar ataxias. Journal of Neurology, 2018, 265, 2040-2051.	3.6	34
21	Predictors of survival in spinocerebellar ataxia type 2 population from Southern Italy. Neurological Sciences, 2018, 39, 1857-1860.	1.9	6
22	The Multiple Faces of Spinocerebellar Ataxia type 2. Annals of Clinical and Translational Neurology, 2017, 4, 687-695.	3.7	24
23	Clinical and genetic characteristics of sporadic adult-onset degenerative ataxia. Neurology, 2017, 89, 1043-1049.	1.1	45
24	Modifications of resting state networks in spinocerebellar ataxia type 2. Movement Disorders, 2015, 30, 1382-1390.	3.9	22
25	Long term follow-up to evaluate the efficacy of miglustat treatment in Italian patients with Niemann-Pick disease type C. Orphanet Journal of Rare Diseases, 2015, 10, 22.	2.7	54
26	Long-term disease progression in spinocerebellar ataxia types 1, 2, 3, and 6: a longitudinal cohort study. Lancet Neurology, The, 2015, 14, 1101-1108.	10.2	213
27	A randomized controlled pilot trial of lithium in spinocerebellar ataxia type 2. Journal of Neurology, 2015, 262, 149-153.	3.6	32
28	Supratentorial and infratentorial damage in spinocerebellar ataxia 2: A diffusionâ€weighted MRI study. Movement Disorders, 2014, 29, 780-786.	3.9	17
29	Prediction of the age at onset in spinocerebellar ataxia type 1, 2, 3 and 6. Journal of Medical Genetics, 2014, 51, 479-486.	3.2	85
30	Mutation of senataxin alters disease-specific transcriptional networks in patients with ataxia with oculomotor apraxia type 2. Human Molecular Genetics, 2014, 23, 4758-4769.	2.9	43
31	Biological and clinical characteristics of individuals at risk for spinocerebellar ataxia types 1, 2, 3, and 6 in the longitudinal RISCA study: analysis of baseline data. Lancet Neurology, The, 2013, 12, 650-658.	10.2	167
32	Reduced cardiac 123I-metaiodobenzylguanidine uptake in patients with spinocerebellar ataxia type 2: a comparative study with Parkinsonâ€™s disease. European Journal of Nuclear Medicine and Molecular Imaging, 2013, 40, 1914-1921.	6.4	16
33	Overview of autosomal recessive ataxias. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2012, 103, 265-274.	1.8	6
34	Depression comorbidity in spinocerebellar ataxia. Movement Disorders, 2011, 26, 870-876.	3.9	69
35	Complex phenotype in an Italian family with a novel mutation in SPG3A. Journal of Neurology, 2010, 257, 328-331.	3.6	5
36	Selfâ€rated health status in spinocerebellar ataxiaâ€”Results from a European multicenter study. Movement Disorders, 2010, 25, 587-595.	3.9	74

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37	Suppression of myoclonus in SCA2 by piracetam. Movement Disorders, 2006, 21, 116-118.	3.9	14
38	Autosomal recessive progressive myoclonus epilepsy with ataxia and mental retardation. Journal of Neurology, 2005, 252, 897-900.	3.6	8
39	The R495W mutation in SPG3A causes spastic paraplegia associated with axonal neuropathy. Journal of Neurology, 2005, 252, 901-903.	3.6	31
40	A pathogenetic classification of hereditary ataxias: Is the time ripe?. Journal of Neurology, 2004, 251, 913-22.	3.6	28
41	Reduced striatal [¹²³I]FPâ€CIT binding in SCA2 patients without parkinsonism. Annals of Neurology, 2004, 55, 426-430.	5.3	49
42	Early-onset inherited ataxias. , 2001, , 519-530.		0
43	Friedreich's ataxia: Point mutations and clinical presentation of compound heterozygotes. Annals of Neurology, 1999, 45, 200-206.	5.3	371
44	Determinants of onset age in Friedreichâ€™s ataxia. Journal of Neurology, 1998, 245, 166-168.	3.6	35
45	Determinants of cognitive disorders in Autosomal Dominant Cerebellar Ataxia type 1. Journal of the Neurological Sciences, 1998, 157, 162-167.	0.6	15
46	May age onset be relevant in the occurrence of left ventricular hypertrophy in Friedreich's ataxia?. Clinical Cardiology, 1997, 20, 141-145.	1.8	19
47	Early-onset ataxia with cardiomyopathy and retained tendon reflexes maps to the friedreich's ataxia locus on chromosome 9q. Annals of Neurology, 1995, 37, 359-362.	5.3	83
48	Prevalence of hereditary ataxias and spastic paraplegias in Molise, a region of Italy. Journal of Neurology, 1992, 239, 351-353.	3.6	106
49	Intrafamilial phenotype variation in Friedreich's disease: possible exceptions to diagnostic criteria. Journal of Neurology, 1991, 238, 147-150.	3.6	17
50	Electrophysiological and histological follow-up study in 15 Friedreich's ataxia patients. Muscle and Nerve, 1990, 13, 536-540.	2.2	37
51	Friedreich's ataxia: electrophysiological and histological findings. Acta Neurologica Scandinavica, 1983, 67, 26-40.	2.1	44