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	Friedreich's ataxia: Point mutations and clinical presentation of compound heterozygotes. Annals of Neurology, 1999, 45, 200-206.	5.3	371
2	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
3	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
4	Prevalence of hereditary ataxias and spastic paraplegias in Molise, a region of Italy. Journal of Neurology, 1992, 239, 351-353.	3.6	106
5	Natural History, Phenotypic Spectrum, and Discriminative Features of Multisystemic RFC1 Disease. Neurology, 2021, 96, e1369-e1382.	1.1	93
6	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
7	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
8	Selfâ€rated health status in spinocerebellar ataxiaâ€"Results from a European multicenter study. Movement Disorders, 2010, 25, 587-595.	3.9	74
9	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
10	Depression comorbidity in spinocerebellar ataxia. Movement Disorders, 2011, 26, 870-876.	3.9	69
11	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
12	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
13	Reduced striatal [¹²³I]FPâ€CIT binding in SCA2 patients without parkinsonism. Annals of Neurology, 2004, 55, 426-430.	5.3	49
14	Clinical and genetic characteristics of sporadic adult-onset degenerative ataxia. Neurology, 2017, 89, 1043-1049.	1.1	45
15	Friedreich's ataxia: electrophysiological and histological findings. Acta Neurologica Scandinavica, 1983, 67, 26-40.	2.1	44
16	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
17	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
18	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

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19	Electrophysiological and histological follow-up study in 15 Friedreich's ataxia patients. Muscle and Nerve, 1990, 13, 536-540.	2.2	37
20	Determinants of onset age in Friedreichâ€™s ataxia. Journal of Neurology, 1998, 245, 166-168.	3.6	35
21	Long-term evolution of patient-reported outcome measures in spinocerebellar ataxias. Journal of Neurology, 2018, 265, 2040-2051.	3.6	34
22	A randomized controlled pilot trial of lithium in spinocerebellar ataxia type 2. Journal of Neurology, 2015, 262, 149-153.	3.6	32
23	The R495W mutation in SPG3A causes spastic paraplegia associated with axonal neuropathy. Journal of Neurology, 2005, 252, 901-903.	3.6	31
24	Spinocerebellar ataxia type 48: last but not least. Neurological Sciences, 2020, 41, 2423-2432.	1.9	31
25	A pathogenetic classification of hereditary ataxias: Is the time ripe?. Journal of Neurology, 2004, 251, 913-22.	3.6	28
26	Degenerative and acquired sporadic adult onset ataxia. Neurological Sciences, 2019, 40, 1335-1342.	1.9	26
27	The Multiple Faces of Spinocerebellar Ataxia type 2. Annals of Clinical and Translational Neurology, 2017, 4, 687-695.	3.7	24
28	Modifications of resting state networks in spinocerebellar ataxia type 2. Movement Disorders, 2015, 30, 1382-1390.	3.9	22
29	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
30	Intrafamilial phenotype variation in Friedreich's disease: possible exceptions to diagnostic criteria. Journal of Neurology, 1991, 238, 147-150.	3.6	17
31	Supratentorial and infratentorial damage in spinocerebellar ataxia 2: A diffusionâ€weighted MRI study. Movement Disorders, 2014, 29, 780-786.	3.9	17
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	Determinants of cognitive disorders in Autosomal Dominant Cerebellar Ataxia type 1. Journal of the Neurological Sciences, 1998, 157, 162-167.	0.6	15
34	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
35	Levels of Neurofilament Light at the Preataxic and Ataxic Stages of Spinocerebellar Ataxia Type 1. Neurology, 2022, 98, .	1.1	15
36	Suppression of myoclonus in SCA2 by piracetam. Movement Disorders, 2006, 21, 116-118.	3.9	14

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37	Prediction of Survival With Longâ€Term Disease Progression in Most Common Spinocerebellar Ataxia. Movement Disorders, 2019, 34, 1220-1227.	3.9	14
38	Spinocerebellar ataxia type 2â€”neuronopathy or neuropathy?. Muscle and Nerve, 2019, 60, 271-278.	2.2	14
39	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
40	NGS in Hereditary Ataxia: When Rare Becomes Frequent. International Journal of Molecular Sciences, 2021, 22, 8490.	4.1	12
41	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
42	Variants in ATP6V0A1 cause progressive myoclonus epilepsy and developmental and epileptic encephalopathy. Brain Communications, 2021, 3, fcab245.	3.3	10
43	Autosomal recessive progressive myoclonus epilepsy with ataxia and mental retardation. Journal of Neurology, 2005, 252, 897-900.	3.6	8
44	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
45	Ataxia-myoclonus syndrome due to a novel homozygous ATP13A2 mutation. Parkinsonism and Related Disorders, 2020, 76, 42-43.	2.2	7
46	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
47	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
48	Predictors of survival in spinocerebellar ataxia type 2 population from Southern Italy. Neurological Sciences, 2018, 39, 1857-1860.	1.9	6
49	Complex phenotype in an Italian family with a novel mutation in SPG3A. Journal of Neurology, 2010, 257, 328-331.	3.6	5
50	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
51	Early-onset inherited ataxias. , 2001, , 519-530.		0