Lorenzo Nanetti

List of Publications by Year
in descending order

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47 papers	1,624 citations	394421 19 h-index	315739 38 g-index
48 all docs	48 docs citations	48 times ranked	2479 citing authors

#	Article	IF	Citations
1	Spinocerebellar Ataxia Type 1: One-Year Longitudinal Study to Identify Clinical and MRI Measures of Disease Progression in Patients and Presymptomatic Carriers. Cerebellum, 2022, 21, 133-144.	2.5	13
2	Digenic inheritance of STUB1 variants and TBP polyglutamine expansions explains the incomplete penetrance of SCA17 and SCA48. Genetics in Medicine, 2022, 24, 29-40.	2.4	24
3	Progression characteristics of the European Friedreich's Ataxia Consortium for Translational Studies (EFACTS): a 4-year cohort study. Lancet Neurology, The, 2021, 20, 362-372.	10.2	53
4	Spastic paraplegia type 46: novel and recurrent GBA2 gene variants in a compound heterozygous Italian patient with spastic ataxia phenotype. Neurological Sciences, 2021, 42, 4741-4745.	1.9	4
5	Late-onset Huntingtonâ€™s disease with 40â€“42 CAG expansion. Neurological Sciences, 2020, 41, 869-876.	1.9	15
6	Huntingtin gene CAG repeat size affects autism risk: Familyâ€based and caseâ€“control association study. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2020, 183, 341-351.	1.7	5
7	Fiberoptic endoscopic evaluation of swallowing in early-to-advanced stage Huntingtonâ€™s disease. Scientific Reports, 2020, 10, 15242.	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	Missing the pathological expansion in Huntington disease: de novo c. 51C >G variant on the expanded allele causing intrafamilial allele dropout. American Journal of Medical Genetics, Part A, 2020, 185, 397-400.	1.2	1
10	In vitro dexamethasone treatment does not induce alternative ATM transcripts in cells from Ataxiaâ€“Telangiectasia patients. Scientific Reports, 2020, 10, 20182.	3.3	3
11	Cortical network dysfunction revealed by magnetoencephalography in carriers of spinocerebellar ataxia 1 or 2 mutation. Clinical Neurophysiology, 2020, 131, 1548-1555.	1.5	4
12	Frequency and distribution of polyQ disease intermediate-length repeat alleles in healthy Italian population. Neurological Sciences, 2020, 41, 1475-1482.	1.9	10
13	Progression of Cerebellar Atrophy in Spinocerebellar Ataxia Type 2 Gene Carriers: A Longitudinal MRI Study in Preclinical and Early Disease Stages. Frontiers in Neurology, 2020, 11, 616419.	2.4	16
14	From congenital microcephaly to adult onset cerebellar ataxia: Distinct and overlapping phenotypes in patients with PNKP gene mutations. American Journal of Medical Genetics, Part A, 2019, 179, 2277-2283.	1.2	18
15	Prediction of Survival With Longâ€Term Disease Progression in Most Common Spinocerebellar Ataxia. Movement Disorders, 2019, 34, 1220-1227.	3.9	14
16	Homozygous variant in OTX2 and possible genetic modifiers identified in a patient with combined pituitary hormone deficiency, ocular involvement, myopathy, ataxia, and mitochondrial impairment. American Journal of Medical Genetics, Part A, 2019, 179, 827-831.	1.2	4
17	ANO10 mutational screening in recessive ataxia: genetic findings and refinement of the clinical phenotype. Journal of Neurology, 2019, 266, 378-385.	3.6	22
18	Multiple system atrophy and CAG repeat length: A genetic screening of polyglutamine disease genes in Italian patients. Neuroscience Letters, 2018, 678, 37-42.	2.1	10

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19	Cortical thickness, stance control, and arithmetic skill: An exploratory study in premanifest Huntington disease. Parkinsonism and Related Disorders, 2018, 51, 17-23.	2.2	22
20	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
21	Long-term evolution of patient-reported outcome measures in spinocerebellar ataxias. Journal of Neurology, 2018, 265, 2040-2051.	3.6	34
22	A recessive ataxia diagnosis algorithm for the next generation sequencing era. Annals of Neurology, 2017, 82, 892-899.	5.3	27
23	Body Mass Index Decline Is Related to Spinocerebellar Ataxia Disease Progression. Movement Disorders Clinical Practice, 2017, 4, 689-697.	1.5	25
24	Stance instability in preclinical SCA1 mutation carriers: A 4-year prospective posturography study. Gait and Posture, 2017, 57, 11-14.	1.4	11
25	Monitoring disease progression in spinocerebellar ataxias: implications for treatment and clinical research. Expert Review of Neurotherapeutics, 2017, 17, 919-931.	2.8	8
26	Cerebellar Involvement in Patients with Mild to Moderate Myoclonus Due to EPM1: Structural and Functional MRI Findings in Comparison with Healthy Controls and Ataxic Patients. Brain Topography, 2017, 30, 380-389.	1.8	5
27	MRI Evidence of Cerebellar and Extraocular Muscle Atrophy Differently Contributing to Eye Movement Abnormalities in SCA2 and SCA28 Diseases. , 2016, 57, 2714.		11
28	SYNE1 ataxia is a common recessive ataxia with major non-cerebellar features: a large multi-centre study. Brain, 2016, 139, 1378-1393.	7.6	87
29	PEX7Mutations Cause Congenital Cataract Retinopathy and Late-Onset Ataxia and Cognitive		

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37	Frontal cortex BOLD signal changes in premanifest Huntington disease. Neurology, 2014, 83, 65-72.	1.1	10
38	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
39	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
40	SETX mutations are a frequent genetic cause of juvenile and adult onset cerebellar ataxia with neuropathy and elevated serum alpha-fetoprotein. Orphanet Journal of Rare Diseases, 2013, 8, 123.	2.7	31
41	ATAXIN2 CAG-repeat length in Italian patients with amyotrophic lateral sclerosis: risk factor or variant phenotype? Implication for genetic testing and counseling. Neurobiology of Aging, 2012, 33, 1847.e15-1847.e21.	3.1	27
42	Erythropoietin in Friedreich ataxia: No effect on frataxin in a randomized controlled trial. Movement Disorders, 2012, 27, 446-449.	3.9	57
43	Lower limb areflexia without central and peripheral conduction abnormalities is highly suggestive of Gerstmannâ€“StrÃ¤usslerâ€“Scheinker disease Pro102Leu. Journal of the Neurological Sciences, 2011, 302, 85-88.	0.6	6
44	Co-occurrence of amyotrophic lateral sclerosis and Charcot-Marie-Tooth disease type 2A in a patient with a novel mutation in the mitofusin-2 gene. Neuromuscular Disorders, 2011, 21, 129-131.	0.6	19
45	Choice-option evaluation is preserved in early Huntington and Parkinsonâ€™s disease. NeuroReport, 2011, 22, 753-757.	1.2	13
46	Slowly progressive sensory hemisyndrome: unusual presentation of paraneoplastic sensory neuronopathy. Journal of the Peripheral Nervous System, 2010, 15, 73-74.	3.1	2
47	Rare association of motor neuron disease and spinocerebellar ataxia type 2 (SCA2): a new case and review of the literature. Journal of Neurology, 2009, 256, 1926-1928.	3.6	42