Alessio Di Fonzo

List of Publications by Year
in descending order

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116	8,140	94433	51608
papers	citations	37	86
		h-index	g-index

121	121	121	10049
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Guidelines for the use and interpretation of assays for monitoring autophagy (4th) Tj ETQq1 1 0.784314 rgBT /Overlock 10 Tf 50 742 Td	9.1	1,430
2	Phenotype, genotype, and worldwide genetic penetrance of LRRK2-associated Parkinson's disease: a case-control study. Lancet Neurology, The, 2008, 7, 583-590.	10.2	1,340
3	A frequent LRRK2 gene mutation associated with autosomal dominant Parkinson's disease. Lancet, The, 2005, 365, 412-415.	13.7	449
4	FBXO7 mutations cause autosomal recessive, early-onset parkinsonian-pyramidal syndrome. Neurology, 2009, 72, 240-245.	1.1	314
5	ATP13A2 missense mutations in juvenile parkinsonism and young onset Parkinson disease. Neurology, 2007, 68, 1557-1562.	1.1	312
6	Mutation in theSYNJ1Gene Associated with Autosomal Recessive, Early-Onset Parkinsonism. Human Mutation, 2013, 34, 1208-1215.	2.5	276
7	A common missense variant in the LRRK2 gene, Gly2385Arg, associated with Parkinsonâ€™s disease risk in Taiwan. Neurogenetics, 2006, 7, 133-138.	1.4	255
8	Adaptive deep brain stimulation in a freely moving parkinsonian patient. Movement Disorders, 2015, 30, 1003-1005.	3.9	198
9	GBA, Gaucher Disease, and Parkinsonâ€™s Disease: From Genetic to Clinic to New Therapeutic Approaches. Cells, 2019, 8, 364.	4.1	187
10	The Role of Mitochondria in Neurodegenerative Diseases: the Lesson from Alzheimerâ€™s Disease and Parkinsonâ€™s Disease. Molecular Neurobiology, 2020, 57, 2959-2980.	4.0	180
11	The G6055A (G2019S) mutation in LRRK2 is frequent in both early and late onset Parkinson's disease and originates from a common ancestor. Journal of Medical Genetics, 2005, 42, e65-e65.	3.2	178
12	The LRRK2 Gly2385Arg variant is associated with Parkinsonâ€™s disease: genetic and functional evidence. Human Genetics, 2007, 120, 857-863.	3.8	157
13	Comprehensive analysis of the LRRK2 gene in sixty families with Parkinson's disease. European Journal of Human Genetics, 2006, 14, 322-331.	2.8	152
14	The Mitochondrial Disulfide Relay System Protein GFER Is Mutated in Autosomal-Recessive Myopathy with Cataract and Combined Respiratory-Chain Deficiency. American Journal of Human Genetics, 2009, 84, 594-604.	6.2	121
15	Mutations in DNA2 Link Progressive Myopathy to Mitochondrial DNA Instability. American Journal of Human Genetics, 2013, 92, 293-300.	6.2	115
16	Cerebellar tDCS: How to Do It. Cerebellum, 2015, 14, 27-30.	2.5	114
17	LRRK2 G2019S mutation and Parkinson's disease: A clinical, neuropsychological and neuropsychiatric study in a large Italian sample. Parkinsonism and Related Disorders, 2006, 12, 410-419.	2.2	106
18	LRP10 genetic variants in familial Parkinson's disease and dementia with Lewy bodies: a genome-wide linkage and sequencing study. Lancet Neurology, The, 2018, 17, 597-608.	10.2	101

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19	POLG mutations in sporadic mitochondrial disorders with multiple mtDNA deletions. Human Mutation, 2003, 22, 498-499.	2.5	100
20	Cerebellar and Motor Cortical Transcranial Stimulation Decrease Levodopa-Induced Dyskinesias in Parkinsonâ€™s Disease. Cerebellum, 2016, 15, 43-47.	2.5	99
21	<scp>A</scp>daptive deep brain stimulation controls levodopaâ€induced side effects in <scp>P</scp>arkinsonian patients. Movement Disorders, 2017, 32, 628-629.	3.9	96
22	Novel ATP13A2 (PARK9) homozygous mutation in a family with marked phenotype variability. Neurogenetics, 2011, 12, 33-39.	1.4	84
23	<scp>GBAâ€Related</scp> Parkinson's Disease: Dissection of Genotypeâ€“Phenotype Correlates in a Large Italian Cohort. Movement Disorders, 2020, 35, 2106-2111.	3.9	83
24	High prevalence ofLRRK2 mutations in familial and sporadic Parkinson's disease in Portugal. Movement Disorders, 2007, 22, 1194-1201.	3.9	76
25	The LRRK2 I2012T, G2019S, and I2020T mutations are rare in Taiwanese patients with sporadic Parkinson's disease. Parkinsonism and Related Disorders, 2005, 11, 521-522.	2.2	70
26	Leucine-Rich Repeat Kinase (LRRK2) Genetics and Parkinsonâ€™s Disease. Advances in Neurobiology, 2017, 14, 3-30.	1.8	66
27	The GBAP1 pseudogene acts as a ceRNA for the glucocerebrosidase gene GBA by sponging miR-22-3p. Scientific Reports, 2017, 7, 12702.	3.3	62
28	The LRRK2 Arg1628Pro variant is a risk factor for Parkinsonâ€™s disease in the Chinese population. Neurogenetics, 2008, 9, 271-276.	1.4	61
29	Remarkable infidelity of polymerase Î³A associated with mutations in POLG1 exonuclease domain. Neurology, 2003, 61, 903-908.	1.1	60
30	Novel mitochondrial protein interactors of immunoglobulin light chains causing heart amyloidosis. FASEB Journal, 2015, 29, 4614-4628.	0.5	60
31	Understanding the pathogenesis of multiple system atrophy: state of the art and future perspectives. Acta Neuropathologica Communications, 2019, 7, 113.	5.2	56
32	Neuropathology of Parkinson's disease associated with theLRRK2 Ile1371Val mutation. Movement Disorders, 2007, 22, 275-278.	3.9	46
33	Mitochondrial Dysregulation and Impaired Autophagy in iPSC-Derived Dopaminergic Neurons of Multiple System Atrophy. Stem Cell Reports, 2018, 11, 1185-1198.	4.8	46
34	Autophagy in motor neuron disease: Key pathogenetic mechanisms and therapeutic targets. Molecular and Cellular Neurosciences, 2016, 72, 84-90.	2.2	43
35	X-linked Parkinsonism with Intellectual Disability caused by novel mutations and somatic mosaicism in RAB39B gene. Parkinsonism and Related Disorders, 2017, 44, 142-146.	2.2	43
36	Striatal dopamine transporter binding in Parkinson's disease associated with theLRRK2 Gly2019Ser mutation. Movement Disorders, 2006, 21, 1144-1147.	3.9	41

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37	SPG11: a consistent clinical phenotype in a family with homozygous Spatacsin truncating mutation. Neurogenetics, 2007, 8, 301-305.	1.4	38
38	Motor and cognitive outcomes of cerebello-spinal stimulation in neurodegenerative ataxia. Brain, 2021, 144, 2310-2321.	7.6	38
39	The SPID-GBA study. Neurology: Genetics, 2020, 6, e523.	1.9	37
40	Nucleoâ€“cytoplasmic transport defects and protein aggregates in neurodegeneration. Translational Neurodegeneration, 2020, 9, 25.	8.0	33
41	Novel missense mutation and large deletion ofGNE gene in autosomal-recessive inclusion-body myopathy. Muscle and Nerve, 2003, 28, 113-117.	2.2	32
42	Mitochondrial dysfunction in fibroblasts of Multiple System Atrophy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 3588-3597.	3.8	32
43	Spinal direct current stimulation (tsDCS) in hereditary spastic paraplegias (HSP): A sham-controlled crossover study. Journal of Spinal Cord Medicine, 2021, 44, 46-53.	1.4	29
44	Discrimination of MSA-P and MSA-C by RT-QuIC analysis of olfactory mucosa: the first assessment of assay reproducibility between two specialized laboratories. Molecular Neurodegeneration, 2021, 16, 82.	10.8	28
45	Genetics of Movement Disorders and the Practicing Clinician; Who and What to Test for?. Current Neurology and Neuroscience Reports, 2018, 18, 37.	4.2	27
46	Mutational analysis of COQ2 in patients with MSA in Italy. Neurobiology of Aging, 2016, 45, 213.e1-213.e2.	3.1	25
47	Neurofascin (NFASC) gene mutation causes autosomal recessive ataxia with demyelinating neuropathy. Parkinsonism and Related Disorders, 2019, 63, 66-72.	2.2	25
48	Obesity and Headache/Migraine: The Importance of Weight Reduction through Lifestyle Modifications. BioMed Research International, 2014, 2014, 1-7.	1.9	24
49	GIGYF2 mutations are not a frequent cause of familial Parkinson's disease. Parkinsonism and Related Disorders, 2009, 15, 703-705.	2.2	22
50	A novel homozygous PLA2G6 mutation causes dystonia-parkinsonism. Parkinsonism and Related Disorders, 2015, 21, 337-339.	2.2	22
51	Real life evaluation of safinamide effectiveness in Parkinsonâ€™s disease. Neurological Sciences, 2018, 39, 733-739.	1.9	22
52	LRRK2 mutations and Parkinson's disease in Sardiniaâ€”A Mediterranean genetic isolate. Parkinsonism and Related Disorders, 2007, 13, 17-21.	2.2	21
53	Lower motor neuron disease with respiratory failure caused by a novel MAPT mutation. Neurology, 2014, 82, 1990-1998.	1.1	21
54	The Length of SNCA Rep1 Microsatellite May Influence Cognitive Evolution in Parkinsonâ€™s Disease. Frontiers in Neurology, 2018, 9, 213.	2.4	21

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55	HOPS-associated neurological disorders (HOPSANDs): linking endolysosomal dysfunction to the pathogenesis of dystonia. Brain, 2021, 144, 2610-2615.	7.6	20
56	Unusual adult-onset Leigh syndrome presentation due to the mitochondrial m.9176T>C mutation. Biochemical and Biophysical Research Communications, 2011, 412, 245-248.	2.1	19
57	Targeting the Autonomic Nervous System for Risk Stratification, Outcome Prediction and Neuromodulation in Ischemic Stroke. International Journal of Molecular Sciences, 2021, 22, 2357.	4.1	19
58	A Practical Approach to Early-Onset Parkinsonism. Journal of Parkinson's Disease, 2022, 12, 1-26.	2.8	19
59	Progressive Encephalomyelitis with Rigidity and Myoclonus Associated With Anti-GlyR Antibodies and Hodgkinâ€™s Lymphoma: A Case Report. Frontiers in Neurology, 2017, 8, 401.	2.4	17
60	Role of Lysosomal Gene Variants in Modulating <scp>GBA</scp>â€Associated Parkinson's Disease Risk. Movement Disorders, 2022, 37, 1202-1210.	3.9	17
61	SLC25A46 mutations in patients with Parkinson's Disease and optic atrophy. Parkinsonism and Related Disorders, 2020, 74, 1-5.	2.2	16
62	A case of CPT deficiency, homoplasmic mtDNA mutation and ragged red fibers at muscle biopsy. Journal of the Neurological Sciences, 2005, 239, 21-24.	0.6	15
63	Parkin polymorphisms and environmental exposure: Decrease in age at onset of Parkinson's disease. NeuroToxicology, 2007, 28, 698-701.	3.0	15
64	A de novo C19orf12 heterozygous mutation in a patient with MPAN. Parkinsonism and Related Disorders, 2018, 48, 109-111.	2.2	15
65	Leukoencephalopathy with calcifications and cysts: Genetic and phenotypic spectrum. American Journal of Medical Genetics, Part A, 2021, 185, 15-25.	1.2	15
66	Abnormal brain temperature in early-onset Parkinson's disease. Movement Disorders, 2016, 31, 425-426.	3.9	14
67	Congenital Myasthenic Syndrome Due to Choline Acetyltransferase Mutations in Infants. Journal of Child Neurology, 2014, 29, 389-393.	1.4	13
68	Pharmacological antagonism of kainate receptor rescues dysfunction and loss of dopamine neurons in a mouse model of human parkin-induced toxicity. Cell Death and Disease, 2020, 11, 963.	6.3	13
69	A Novel Homozygous <scp>VPS11</scp> Variant May Cause Generalized Dystonia. Annals of Neurology, 2021, 89, 834-839.	5.3	13
70	Genetic variants in levodopa-induced dyskinesia (LID): A systematic review and meta-analysis. Parkinsonism and Related Disorders, 2021, 84, 52-60.	2.2	13
71	Autosomal dominant restless legs syndrome maps to chromosome 20p13 (RLSâ€5) in a Dutch kindred. Movement Disorders, 2010, 25, 1715-1722.	3.9	12
72	The Italian tremor Network (TITAN): rationale, design and preliminary findings. Neurological Sciences, 2022, 43, 5369-5376.	1.9	12

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73	Mutations in TMEM230 are rare in autosomal dominant Parkinson's disease. Parkinsonism and Related Disorders, 2017, 39, 87-88.	2.2	11
74	In vitro models of multiple system atrophy from primary cells to induced pluripotent stem cells. Journal of Cellular and Molecular Medicine, 2018, 22, 2536-2546.	3.6	11
75	LRRK2 MUTATION ANALYSIS IN PARKINSON DISEASE FAMILIES WITH EVIDENCE OF LINKAGE TO PARK8. Neurology, 2008, 70, 2348-2349.	1.1	10
76	Pseudoâ€orthostatic and resting leg tremor in a large spanish family with homozygous truncating parkin mutation. Movement Disorders, 2009, 24, 144-147.	3.9	10
77	Juvenile dystonia-parkinsonism syndrome caused by a novel p.S941Tfs1X ATP13A2 (PARK9) mutation. Parkinsonism and Related Disorders, 2015, 21, 1378-1380.	2.2	10
78	Dystoniaâ€ataxia syndrome with permanent torsional nystagmus caused by ECHS1 deficiency. Annals of Clinical and Translational Neurology, 2020, 7, 839-845.	3.7	10
79	<scp>TWNK</scp> in Parkinson's Disease: A Movement Disorder and Mitochondrial Disease Center Perspective Study. Movement Disorders, 2022, 37, 1938-1943.	3.9	10
80	Globus pallidus internus deep brain stimulation in PINK-1 related Parkinson's disease: A case report. Parkinsonism and Related Disorders, 2017, 38, 93-94.	2.2	9
81	Loss of the nucleoporin Aladin in central nervous system and fibroblasts of Allgrove Syndrome. Human Molecular Genetics, 2019, 28, 3921-3927.	2.9	9
82	Unravelling Genetic Factors Underlying Corticobasal Syndrome: A Systematic Review. Cells, 2021, 10, 171.	4.1	8
83	VPS13C-associated Parkinson's disease: Two novel cases and review of the literature. Parkinsonism and Related Disorders, 2022, 94, 37-39.	2.2	8
84	Two novel mutations in PEO1 (Twinkle) gene associated with chronic external ophthalmoplegia. Journal of the Neurological Sciences, 2011, 308, 173-176.	0.6	7
85	A rapid and low-cost test for screening the most common Parkinson's disease-related GBA variants. Parkinsonism and Related Disorders, 2020, 80, 138-141.	2.2	7
86	Systemic involvement in adultâ€onset leukoencephalopathy with intracranial calcifications and cysts (Labrune syndrome) with a novel mutation of the SNORD118 gene. European Journal of Neurology, 2020, 27, 2329-2332.	3.3	6
87	Dysautonomia in Parkinsonâ€™s Disease: Impact of Glucocerebrosidase Gene Mutations on Cardiovascular Autonomic Control. Frontiers in Neuroscience, 2022, 16, 842498.	2.8	6
88	Validation of the Italian version of the PSP Quality of Life questionnaire. Neurological Sciences, 2019, 40, 2587-2594.	1.9	5
89	Validation of the Italian version of carersâ€™ quality-of-life questionnaire for parkinsonism (PQoL) Tj ETQq1 1 0.784314 rgBT /Overlock	1.9	5
90	Screening of LRP10 mutations in Parkinson's disease patients from Italy. Parkinsonism and Related Disorders, 2021, 89, 17-21.	2.2	5

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91	The novel mitochondrial tRNAAsn gene mutation m.5709T>C produces ophthalmoparesis and respiratory impairment. European Journal of Human Genetics, 2012, 20, 357-360.	2.8	4
92	Parkinsonism in diseases predominantly presenting with dystonia. International Review of Neurobiology, 2019, 149, 307-326.	2.0	4
93	Progressive myoclonus without epilepsy due to a NUS1 frameshift insertion: Dyssynergia cerebellaris myoclonica revisited. Parkinsonism and Related Disorders, 2022, 98, 53-55.	2.2	4
94	Clinical uses of Bupropion in patients with Parkinsonâ€™s disease and comorbid depressive or neuropsychiatric symptoms: a scoping review. BMC Neurology, 2022, 22, 169.	1.8	4
95	Parkinsonâ€™s disease in Gaucher disease patients: whatâ€™s changing in the counseling and management of patients and their relatives?. Orphanet Journal of Rare Diseases, 2020, 15, 262.	2.7	3
96	Comprehensive Genomic Analysis Reveals the Prognostic Role of LRRK2 Copy-Number Variations in Human Malignancies. Genes, 2020, 11, 846.	2.4	3
97	A case report of late-onset cerebellar ataxia associated with a rare p.R342W TGM6 (SCA35) mutation. BMC Neurology, 2020, 20, 408.	1.8	3
98	Design and Operation of the Lombardy Parkinson's Disease Network. Frontiers in Neurology, 2020, 11, 573.	2.4	3
99	Microscopic Polyangiitis With Selective Involvement of Central and Peripheral Nervous System: A Case Report. Frontiers in Neurology, 2020, 11, 269.	2.4	3
100	Clinical Reasoning: A 75-year-old man with parkinsonism, mood depression, and weight loss. Neurology, 2018, 90, 572-575.	1.1	2
101	Sodium Levels Predict Disability at Discharge in Guillain-BarrÃ© Syndrome: A Retrospective Cohort Study. Frontiers in Neurology, 2021, 12, 729252.	2.4	2
102	Parkinsonism and ataxia. Journal of the Neurological Sciences, 2021, , 120020.	0.6	2
103	Childhoodâ€onset dystonia with cerebellar signs: expanding the spectrum of GNAL mutations. European Journal of Neurology, 2020, 27, e66-e67.	3.3	1
104	Transcriptomic characterization of tissues from patients and subsequent pathway analyses reveal biological pathways that are implicated in spastic ataxia. Cell and Bioscience, 2022, 12, 29.	4.8	1
105	1.283 Clinical and genetic study of a large Dutch family with autosomal dominant restless legs syndrome. Parkinsonism and Related Disorders, 2007, 13, S72.	2.2	0
106	2.119 Parkin polymorphisms and environmental exposure: Reduction of Parkinson's disease age of onset. Parkinsonism and Related Disorders, 2007, 13, S97.	2.2	0
107	Generation and characterization of iPSC-derived cortical pyramidal neurons from patients affected by multiple system atrophy. Parkinsonism and Related Disorders, 2016, 22, e119-e120.	2.2	0
108	LRRK2 and GBA mutation analysis in a cohort of Italian familial and sporadic PD. Parkinsonism and Related Disorders, 2016, 22, e167.	2.2	0

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109	Hallucinations in Neurological Disorders. , 2018, , 99-130.		0
110	Syncope and autonomic failure in a middle-aged man. Internal and Emergency Medicine, 2019, 14, 271-274.	2.0	0
111	Late-onset leukoencephalopathy in a patient with recessive EARS2 mutations. Neurology: Genetics, 2020, 6, e488.	1.9	0
112	Expanding the genotypic and phenotypic spectrum of Betaâ€propeller poteinâ€associated neurodegeneration. European Journal of Neurology, 2021, 28, e25-e27.	3.3	0
113	Clinical features and disease course of patients with acute ischaemic stroke just before the Italian index case: Was COVID-19 already there?. Internal and Emergency Medicine, 2021, 16, 1247-1252.	2.0	0
114	A 79-year-old man with unexplained recurrent syncope and severe orthostatic hypotension. Internal and Emergency Medicine, 2021, , 1.	2.0	0
115	The SPID- <i>GBA</i> Study: The Largest Monocentric Study on Sex Distribution, Penetrance, Incidence, and Association with Dementia of <i>GBA</i> Mutations in Parkinson's Disease. SSRN Electronic Journal, 0, , .	0.4	0
116	Genetic evaluation in phenotypically discordant monozygotic twins with Coats Disease. European Journal of Ophthalmology, 0, , 112067212211077.	1.3	0