Niccolo E Mencacci

List of Publications by Year
in descending order

82	5,645	117625 34	95266 68
papers	citations	h-index	g-index

87	87	87	7791
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Identification of novel risk loci, causal insights, and heritable risk for Parkinson's disease: a meta-analysis of genome-wide association studies. Lancet Neurology, The, 2019, 18, 1091-1102.	10.2	1,414
2	Glucocerebrosidase mutations influence the natural history of Parkinsonâ€™s disease in a community-based incident cohort. Brain, 2013, 136, 392-399.	7.6	266
3	Long-term Clinical Outcome of Fetal Cell Transplantation for Parkinson Disease. JAMA Neurology, 2014, 71, 83.	9.0	257
4	Dystonia. Nature Reviews Disease Primers, 2018, 4, 25.	30.5	223
5	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237.	21.4	186
6	Parkinsonâ€™s disease in GTP cyclohydrolase 1 mutation carriers. Brain, 2014, 137, 2480-2492.	7.6	169
7	Mitofusin-mediated ER stress triggers neurodegeneration in pink1/parkin models of Parkinsonâ€™s disease. Cell Death and Disease, 2016, 7, e2271-e2271.	6.3	151
8	Mutations in the autoregulatory domain of Î²â€tubulin 4a cause hereditary dystonia. Annals of Neurology, 2013, 73, 546-553.	5.3	148
9	ADCY5 mutations are another cause of benign hereditary chorea. Neurology, 2015, 85, 80-88.	1.1	140
10	A genome-wide association study in multiple system atrophy. Neurology, 2016, 87, 1591-1598.	1.1	139
11	The glucocerobrosidase E326K variant predisposes to Parkinson's disease, but does not cause Gaucher's disease. Movement Disorders, 2013, 28, 232-236.	3.9	121
12	Parkin Disease. JAMA Neurology, 2013, 70, 571.	9.0	119
13	Hâ€ABC syndrome and DYT4: Variable expressivity or pleiotropy of TUBB4 mutations?. Movement Disorders, 2015, 30, 828-833.	3.9	117
14	A Missense Mutation in KCTD17 Causes Autosomal Dominant Myoclonus-Dystonia. American Journal of Human Genetics, 2015, 96, 938-947.	6.2	109
15	Recessive mutations in VPS13D cause childhood onset movement disorders. Annals of Neurology, 2018, 83, 1089-1095.	5.3	104
16	De Novo Mutations in PDE10A Cause Childhood-Onset Chorea with Bilateral Striatal Lesions. American Journal of Human Genetics, 2016, 98, 763-771.	6.2	96
17	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. JAMA Neurology, 2021, 78, 464.	9.0	95
18	Cortical pencil lining in neuroferritinopathy: A diagnostic clue. Neurology, 2015, 84, 1816-1818.	1.1	93

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19	Genotype and phenotype in Parkinson's disease: Lessons in heterogeneity from deep brain stimulation. Movement Disorders, 2013, 28, 1370-1375.	3.9	77
20	Deletions at 22q11.2 in idiopathic Parkinson's disease: a combined analysis of genome-wide association data. Lancet Neurology, The, 2016, 15, 585-596.	10.2	77
21	Lossâ€ofâ€Function Variants in <scp>HOPS</scp> Complex Genes <scp>VPS16</scp> and <scp>VPS41</scp> Cause Early Onset Dystonia Associated with Lysosomal Abnormalities. Annals of Neurology, 2020, 88, 867-877.	5.3	70
22	ADCY5-related movement disorders: Frequency, disease course and phenotypic variability in a cohort of paediatric patients. Parkinsonism and Related Disorders, 2017, 41, 37-43.	2.2	67
23	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. Movement Disorders, 2019, 34, 460-468.	3.9	66
24	PREDICT-PD: An online approach to prospectively identify risk indicators of Parkinson's disease. Movement Disorders, 2017, 32, 219-226.	3.9	59
25	Penetrance of Parkinson's Disease in LRRK2 p.G2019S Carriers Is Modified by a Polygenic Risk Score. Movement Disorders, 2020, 35, 774-780.	3.9	57
26	Dopaminergic Neuronal Imaging in Genetic Parkinson's Disease: Insights into Pathogenesis. PLoS ONE, 2013, 8, e69190.	2.5	55
27	Frequency and phenotypic spectrum of KMT2B dystonia in childhood: A singleâ€center cohort study. Movement Disorders, 2019, 34, 1516-1527.	3.9	55
28	Low anaerobic threshold and increased skeletal muscle lactate production in subjects with Huntington's disease. Movement Disorders, 2011, 26, 130-137.	3.9	52
29	A homozygous lossâ€ofâ€function mutation in PDE2A associated to earlyâ€onset hereditary chorea. Movement Disorders, 2018, 33, 482-488.	3.9	52
30	Emerging Monogenic Complex Hyperkinetic Disorders. Current Neurology and Neuroscience Reports, 2017, 17, 97.	4.2	51
31	Dystonia genes functionally converge in specific neurons and share neurobiology with psychiatric disorders. Brain, 2020, 143, 2771-2787.	7.6	50
32	SLC25A46 mutations underlie progressive myoclonic ataxia with optic atrophy and neuropathy. Movement Disorders, 2016, 31, 1249-1251.	3.9	49
33	The Genetic Architecture of Parkinson Disease in Spain: Characterizing Populationâ€Specific Risk, Differential Haplotype Structures, and Providing Etiologic Insight. Movement Disorders, 2019, 34, 1851-1863.	3.9	47
34	Myoclonus-dystonia syndrome due to tyrosine hydroxylase deficiency. Neurology, 2012, 79, 435-441.	1.1	45
35	GBA-Associated Parkinsonâ€™s Disease: Progression in a Deep Brain Stimulation Cohort. Journal of Parkinson's Disease, 2017, 7, 635-644.	2.8	44
36	Finding genetically-supported drug targets for Parkinsonâ€™s disease using Mendelian randomization of the druggable genome. Nature Communications, 2021, 12, 7342.	12.8	44

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37	All in the blink of an eye: new insight into cerebellar and brainstem function in <scp>DYT</scp>1 and <scp>DYT</scp>6 dystonia. European Journal of Neurology, 2015, 22, 762-767.	3.3	38
38	PDE10A and ADCY5 mutations linked to molecular and microstructural basal ganglia pathology. Movement Disorders, 2018, 33, 1961-1965.	3.9	38
39	Partial loss-of-function of sodium channel SCN8A in familial isolated myoclonus. Human Mutation, 2018, 39, 965-969.	2.5	34
40	Aripiprazole in the treatment of Huntington's disease: a case series. Neuropsychiatric Disease and Treatment, 2009, 5, 1-4.	2.2	34
41	<scp>EIF2AK2</scp> Missense Variants Associated with Early Onset Generalized Dystonia. Annals of Neurology, 2021, 89, 485-497.	5.3	32
42	Genetic Dystonias: Update on Classification and New Genetic Discoveries. Current Neurology and Neuroscience Reports, 2021, 21, 8.	4.2	32
43	Metalloproteinase alterations in the bone marrow of ALS patients. Journal of Molecular Medicine, 2010, 88, 553-564.	3.9	30
44	Emerging and converging molecular mechanisms in dystonia. Journal of Neural Transmission, 2021, 128, 483-498.	2.8	29
45	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. Annals of Neurology, 2021, 90, 35-42.	5.3	29
46	TheCACNA1BR1389H variant is not associated with myoclonus-dystonia in a large European multicentric cohort. Human Molecular Genetics, 2015, 24, 5326-5329.	2.9	28
47	Recent advances in genetics of chorea. Current Opinion in Neurology, 2016, 29, 486-495.	3.6	25
48	Oligoclonal bands in the cerebrospinal fluid of amyotrophic lateral sclerosis patients with disease-associated mutations. Journal of Neurology, 2013, 260, 85-92.	3.6	24
49	Analysis of the genetic variability in Parkinson's disease from Southern Spain. Neurobiology of Aging, 2016, 37, 210.e1-210.e5.	3.1	23
50	Expanding the Phenotype and Genetic Defects Associated with the <scp>GOSR</scp>2 Gene. Movement Disorders Clinical Practice, 2015, 2, 271-273.	1.5	21
51	The Emerging Role of Phosphodiesterases in Movement Disorders. Movement Disorders, 2021, 36, 2225-2243.	3.9	21
52	Intracerebral haemorrhage, a possible presentation in Churg-Strauss syndrome: Case report and review of the literature. Journal of the Neurological Sciences, 2011, 301, 107-111.	0.6	19
53	Biallelic variants in TSPOAP1, encoding the active-zone protein RIMBP1, cause autosomal recessive dystonia. Journal of Clinical Investigation, 2021, 131, .	8.2	18
54	<scp>YY1</scp>â€Related Dystonia: Clinical Aspects and Longâ€Term Response to Deep Brain Stimulation. Movement Disorders, 2021, 36, 1461-1462.	3.9	16

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55	The commercial genetic testing landscape for Parkinson's disease. Parkinsonism and Related Disorders, 2021, 92, 107-111.	2.2	16
56	LRP10 in Î±-synucleinopathies. Lancet Neurology, The, 2018, 17, 1032.	10.2	15
57	<scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833.	5.3	14
58	Biallelic <scp>AOPEP</scp> Lossâ€ofâ€Function Variants Cause Progressive Dystonia with Prominent Limb Involvement. Movement Disorders, 2022, 37, 137-147.	3.9	14
59	A PDE10A de novo mutation causes childhoodâ€onset chorea with diurnal fluctuations. Movement Disorders, 2017, 32, 1646-1647.	3.9	13
60	Paroxysmal Kinesigenic Dyskinesia May Be Misdiagnosed in Co-occurring Gilles de la Tourette Syndrome. Movement Disorders Clinical Practice, 2014, 1, 84-86.	1.5	11
61	Oculomotor apraxia and disrupted sleep with nocturnal ballistic bouts in ADCY5-related disease. Parkinsonism and Related Disorders, 2018, 54, 103-106.	2.2	10
62	KCTD17 is a confirmed new gene for dystonia, but is it responsible for SGCE-negative myoclonus-dystonia?. Parkinsonism and Related Disorders, 2019, 61, 1-3.	2.2	10
63	Mitochondrial complex I NUBPL mutations cause combined dystonia with bilateral striatal necrosis and cerebellar atrophy. European Journal of Neurology, 2019, 26, 1240-1243.	3.3	9
64	Persistent chorea in DYT6, due to anticholinergic therapy. Parkinsonism and Related Disorders, 2015, 21, 1282-1283.	2.2	8
65	Ropinirole monotherapy induced severe reversible dyskinesias in Parkinson's disease. Movement Disorders, 2013, 28, 1159-1160.	3.9	6
66	Webâ€based assessment of Parkinson's prodromal markers identifies GBA variants. Movement Disorders, 2015, 30, 1002-1003.	3.9	6
67	Neurodegeneration With Brain Iron Accumulation (NBIA) Syndromes Presenting With Late-Onset Craniocervical Dystonia: An Illustrative Case Seriesâ€Ž. Movement Disorders Clinical Practice, 2017, 4, 254-257.	1.5	6
68	Restless Legs Syndrome in NKX2-1-related chorea: An expansion of the disease spectrum. Brain and Development, 2019, 41, 250-256.	1.1	6
69	Childhoodâ€Onset Chorea Caused by a Recurrent De Novo DRD2 Variant. Movement Disorders, 2021, 36, 1472-1473.	3.9	6
70	Atypical Parkinsonism Revealing a Late Onset, Rigid and Akinetic Form of Huntington's Disease. Case Reports in Neurological Medicine, 2011, 2011, 1-3.	0.4	5
71	Reply: Parkinsonâ€™s disease in GTP cyclohydrolase 1 mutation carriers. Brain, 2015, 138, e352-e352.	7.6	4
72	ADCY5 mutations are another cause of benign hereditary chorea. Neurology, 2016, 86, 978-979.	1.1	4

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73	A Novel SGCE Nonsense Variant Associated With Marked Intrafamilial Variability in a Turkish Family With Myoclonusâ€Dystonia. Movement Disorders Clinical Practice, 2019, 6, 479-482.	1.5	3
74	Replication assessment of NUS1 variants in Parkinson's disease. Neurobiology of Aging, 2021, 101, 300.e1-300.e3.	3.1	3
75	Communityâ€based genetic study of Parkinson's disease in Estonia. Acta Neurologica Scandinavica, 2021, 143, 89-95.	2.1	3
76	Clinical Reasoning: A 77-Year-Old Man With Involuntary Movements, Sleep Changes, Falls, Bulbar Symptoms, and Cognitive Complaints. Neurology, 2022, 99, 26-30.	1.1	2
77	Screening of mutations in NOL3 in a myoclonic syndromes series. Journal of Neurology, 2014, 261, 1830-1831.	3.6	1
78	Dystonia; a roadmap is needed for future genetic studies. Parkinsonism and Related Disorders, 2019, 58, 9-11.	2.2	1
79	The Endless Expansion of the Phenotypic Spectrum of <scp>ATP</scp>1A3 Mutations: A True Diagnostic Challenge. Movement Disorders Clinical Practice, 2016, 3, 395-397.	1.5	0
80	The global Parkinson's Genetics Program (GP2): The Monogenic Hub 500-genomes pilot project. Journal of the Neurological Sciences, 2021, 429, 118257.	0.6	0
81	Monogenic portal of the global Parkinson's Genetics Program (GP2). Journal of the Neurological Sciences, 2021, 429, 118256.	0.6	0
82	Naming Genes for Dystonia: DYT-z or Ditzy?. Tremor and Other Hyperkinetic Movements, 2019, 9, .	2.0	0