# 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 | Biallelic <scp>AOPEP</scp> Lossâ€ofâ€Function Variants Cause Progressive Dystonia with Prominent Limb Involvement. Movement Disorders, 2022, 37, 137-147. | 3.9 | 14 |
| 2 | 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 |
| 3 | <scp>EIF2AK2</scp> Missense Variants Associated with Early Onset Generalized Dystonia. Annals of Neurology, 2021, 89, 485-497. | 5.3 | 32 |
| 4 | Replication assessment of NUS1 variants in Parkinson's disease. Neurobiology of Aging, 2021, 101, 300.e1-300.e3. | 3.1 | 3 |
| 5 | Communityâ€based genetic study of Parkinson's disease in Estonia. Acta Neurologica Scandinavica, 2021, 143, 89-95. | 2.1 | 3 |
| 6 | Emerging and converging molecular mechanisms in dystonia. Journal of Neural Transmission, 2021, 128, 483-498. | 2.8 | 29 |
| 7 | <scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833. | 5.3 | 14 |
| 8 | Genetic Dystonias: Update on Classification and New Genetic Discoveries. Current Neurology and Neuroscience Reports, 2021, 21, 8. | 4.2 | 32 |
| 9 | <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 |
| 10 | Biallelic variants in TSPOAP1, encoding the active-zone protein RIMBP1, cause autosomal recessive dystonia. Journal of Clinical Investigation, 2021, 131, . | 8.2 | 18 |
| 11 | 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 |
| 12 | Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. Annals of Neurology, 2021, 90, 35-42. | 5.3 | 29 |
| 13 | Childhoodâ€Onset Chorea Caused by a Recurrent De Novo DRD2 Variant. Movement Disorders, 2021, 36, 1472-1473. | 3.9 | 6 |
| 14 | The Emerging Role of Phosphodiesterases in Movement Disorders. Movement Disorders, 2021, 36, 2225-2243. | 3.9 | 21 |
| 15 | 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 |
| 16 | Monogenic portal of the global Parkinson's Genetics Program (GP2). Journal of the Neurological Sciences, 2021, 429, 118256. | 0.6 | 0 |
| 17 | The commercial genetic testing landscape for Parkinson's disease. Parkinsonism and Related Disorders, 2021, 92, 107-111. | 2.2 | 16 |
| 18 | Finding genetically-supported drug targets for Parkinsonâ€™s disease using Mendelian randomization of the druggable genome. Nature Communications, 2021, 12, 7342. | 12.8 | 44 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Dystonia genes functionally converge in specific neurons and share neurobiology with psychiatric disorders. Brain, 2020, 143, 2771-2787. | 7.6 | 50 |
| 20 | 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 |
| 21 | 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 |
| 22 | 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 |
| 23 | 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 |
| 24 | 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 |
| 25 | Frequency and phenotypic spectrum of KMT2B dystonia in childhood: A singleâ€center cohort study. Movement Disorders, 2019, 34, 1516-1527. | 3.9 | 55 |
| 26 | 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 |
| 27 | 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 |
| 28 | 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 |
| 29 | Dystonia; a roadmap is needed for future genetic studies. Parkinsonism and Related Disorders, 2019, 58, 9-11. | 2.2 | 1 |
| 30 | Restless Legs Syndrome in NKX2-1-related chorea: An expansion of the disease spectrum. Brain and Development, 2019, 41, 250-256. | 1.1 | 6 |
| 31 | Naming Genes for Dystonia: DYT-z or Ditzy?. Tremor and Other Hyperkinetic Movements, 2019, 9, . | 2.0 | 0 |
| 32 | Recessive mutations in VPS13D cause childhood onset movement disorders. Annals of Neurology, 2018, 83, 1089-1095. | 5.3 | 104 |
| 33 | Oculomotor apraxia and disrupted sleep with nocturnal ballistic bouts in ADCY5-related disease. Parkinsonism and Related Disorders, 2018, 54, 103-106. | 2.2 | 10 |
| 34 | A homozygous lossâ€ofâ€function mutation in PDE2A associated to earlyâ€onset hereditary chorea. Movement Disorders, 2018, 33, 482-488. | 3.9 | 52 |
| 35 | LRP10 in Î±-synucleinopathies. Lancet Neurology, The, 2018, 17, 1032. | 10.2 | 15 |
| 36 | PDE10A and ADCY5 mutations linked to molecular and microstructural basal ganglia pathology. Movement Disorders, 2018, 33, 1961-1965. | 3.9 | 38 |

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|---|---|---|---|
| 37 | Dystonia. Nature Reviews Disease Primers, 2018, 4, 25. | 30.5 | 223 |
| 38 | Partial loss-of-function of sodium channel SCN8A in familial isolated myoclonus. Human Mutation, 2018, 39, 965-969. | 2.5 | 34 |
| 39 | PREDICT-PD: An online approach to prospectively identify risk indicators of Parkinson's disease. Movement Disorders, 2017, 32, 219-226. | 3.9 | 59 |
| 40 | 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 |
| 41 | Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237. | 21.4 | 186 |
| 42 | Emerging Monogenic Complex Hyperkinetic Disorders. Current Neurology and Neuroscience Reports, 2017, 17, 97. | 4.2 | 51 |
| 43 | A PDE10A de novo mutation causes childhoodâ€onset chorea with diurnal fluctuations. Movement Disorders, 2017, 32, 1646-1647. | 3.9 | 13 |
| 44 | GBA-Associated Parkinsonâ€™s Disease: Progression in a Deep Brain Stimulation Cohort. Journal of Parkinson's Disease, 2017, 7, 635-644. | 2.8 | 44 |
| 45 | 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 |
| 46 | 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 |
| 47 | Recent advances in genetics of chorea. Current Opinion in Neurology, 2016, 29, 486-495. | 3.6 | 25 |
| 48 | 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 |
| 49 | A genome-wide association study in multiple system atrophy. Neurology, 2016, 87, 1591-1598. | 1.1 | 139 |
| 50 | SLC25A46 mutations underlie progressive myoclonic ataxia with optic atrophy and neuropathy. Movement Disorders, 2016, 31, 1249-1251. | 3.9 | 49 |
| 51 | 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 |
| 52 | Analysis of the genetic variability in Parkinson's disease from Southern Spain. Neurobiology of Aging, 2016, 37, 210.e1-210.e5. | 3.1 | 23 |
| 53 | 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 |
| 54 | ADCY5 mutations are another cause of benign hereditary chorea. Neurology, 2016, 86, 978-979. | 1.1 | 4 |

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|---|---|---|---|
| 55 | Webâ€based assessment of Parkinson's prodromal markers identifies GBA variants. Movement Disorders, 2015, 30, 1002-1003. | 3.9 | 6 |
| 56 | 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 |
| 57 | A Missense Mutation in KCTD17 Causes Autosomal Dominant Myoclonus-Dystonia. American Journal of Human Genetics, 2015, 96, 938-947. | 6.2 | 109 |
| 58 | Hâ€ABC syndrome and DYT4: Variable expressivity or pleiotropy of TUBB4 mutations?. Movement Disorders, 2015, 30, 828-833. | 3.9 | 117 |
| 59 | Reply: Parkinsonâ€™s disease in GTP cyclohydrolase 1 mutation carriers. Brain, 2015, 138, e352-e352. | 7.6 | 4 |
| 60 | ADCY5 mutations are another cause of benign hereditary chorea. Neurology, 2015, 85, 80-88. | 1.1 | 140 |
| 61 | TheCACNA1BR1389H variant is not associated with myoclonus-dystonia in a large European multicentric cohort. Human Molecular Genetics, 2015, 24, 5326-5329. | 2.9 | 28 |
| 62 | Persistent chorea in DYT6, due to anticholinergic therapy. Parkinsonism and Related Disorders, 2015, 21, 1282-1283. | 2.2 | 8 |
| 63 | Cortical pencil lining in neuroferritinopathy: A diagnostic clue. Neurology, 2015, 84, 1816-1818. | 1.1 | 93 |
| 64 | 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 |
| 65 | Long-term Clinical Outcome of Fetal Cell Transplantation for Parkinson Disease. JAMA Neurology, 2014, 71, 83. | 9.0 | 257 |
| 66 | 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 |
| 67 | Parkinsonâ€™s disease in GTP cyclohydrolase 1 mutation carriers. Brain, 2014, 137, 2480-2492. | 7.6 | 169 |
| 68 | Screening of mutations in NOL3 in a myoclonic syndromes series. Journal of Neurology, 2014, 261, 1830-1831. | 3.6 | 1 |
| 69 | Ropinirole monotherapy induced severe reversible dyskinesias in Parkinson's disease. Movement Disorders, 2013, 28, 1159-1160. | 3.9 | 6 |
| 70 | 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 |
| 71 | Glucocerebrosidase mutations influence the natural history of Parkinsonâ€™s disease in a community-based incident cohort. Brain, 2013, 136, 392-399. | 7.6 | 266 |
| 72 | 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 |

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|---|---|---|---|
| 73 | Parkin Disease. JAMA Neurology, 2013, 70, 571. | 9.0 | 119 |
| 74 | Mutations in the autoregulatory domain of Î²â€tubulin 4a cause hereditary dystonia. Annals of Neurology, 2013, 73, 546-553. | 5.3 | 148 |
| 75 | Genotype and phenotype in Parkinson's disease: Lessons in heterogeneity from deep brain stimulation. Movement Disorders, 2013, 28, 1370-1375. | 3.9 | 77 |
| 76 | Dopaminergic Neuronal Imaging in Genetic Parkinson's Disease: Insights into Pathogenesis. PLoS ONE, 2013, 8, e69190. | 2.5 | 55 |
| 77 | Myoclonus-dystonia syndrome due to tyrosine hydroxylase deficiency. Neurology, 2012, 79, 435-441. | 1.1 | 45 |
| 78 | 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 |
| 79 | 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 |
| 80 | Low anaerobic threshold and increased skeletal muscle lactate production in subjects with Huntington's disease. Movement Disorders, 2011, 26, 130-137. | 3.9 | 52 |
| 81 | Metalloproteinase alterations in the bone marrow of ALS patients. Journal of Molecular Medicine, 2010, 88, 553-564. | 3.9 | 30 |
| 82 | Aripiprazole in the treatment of Huntington's disease: a case series. Neuropsychiatric Disease and Treatment, 2009, 5, 1-4. | 2.2 | 34 |