

Gael Nicolas

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

84
papers

3,327
citations

27
h-index

57
g-index

91
ext. papers

4,878
ext. citations

7.9
avg, IF

4.36
L-index

#	Paper	IF	Citations
84	Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates A β , tau, immunity and lipid processing. <i>Nature Genetics</i> , 2019 , 51, 414-430	36.3	917
83	Mutations in the gene encoding PDGF-B cause brain calcifications in humans and mice. <i>Nature Genetics</i> , 2013 , 45, 1077-82	36.3	214
82	APP, PSEN1, and PSEN2 mutations in early-onset Alzheimer disease: A genetic screening study of familial and sporadic cases. <i>PLoS Medicine</i> , 2017 , 14, e1002270	11.6	204
81	Mutation of the PDGFRB gene as a cause of idiopathic basal ganglia calcification. <i>Neurology</i> , 2013 , 80, 181-7	6.5	192
80	Mutations in XPR1 cause primary familial brain calcification associated with altered phosphate export. <i>Nature Genetics</i> , 2015 , 47, 579-81	36.3	176
79	Phenotypic spectrum of probable and genetically-confirmed idiopathic basal ganglia calcification. <i>Brain</i> , 2013 , 136, 3395-407	11.2	146
78	SQSTM1 mutations in French patients with frontotemporal dementia or frontotemporal dementia with amyotrophic lateral sclerosis. <i>JAMA Neurology</i> , 2013 , 70, 1403-10	17.2	131
77	Whole exome sequencing study identifies novel rare and common Alzheimer's-Associated variants involved in immune response and transcriptional regulation. <i>Molecular Psychiatry</i> , 2020 , 25, 1859-1875	15.1	106
76	Contribution to Alzheimer's disease risk of rare variants in TREM2, SORL1, and ABCA7 in 1779 cases and 1273 controls. <i>Neurobiology of Aging</i> , 2017 , 59, 220.e1-220.e9	5.6	83
75	SORL1 rare variants: a major risk factor for familial early-onset Alzheimer's disease. <i>Molecular Psychiatry</i> , 2016 , 21, 831-6	15.1	71
74	Screening of dementia genes by whole-exome sequencing in early-onset Alzheimer disease: input and lessons. <i>European Journal of Human Genetics</i> , 2016 , 24, 710-6	5.3	61
73	Brain calcification process and phenotypes according to age and sex: Lessons from SLC20A2, PDGFB, and PDGFRB mutation carriers. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2015 , 168, 586-94	3.5	61
72	Update and Mutational Analysis of SLC20A2: A Major Cause of Primary Familial Brain Calcification. <i>Human Mutation</i> , 2015 , 36, 489-95	4.7	54
71	TYROBP genetic variants in early-onset Alzheimer's disease. <i>Neurobiology of Aging</i> , 2016 , 48, 222.e9-222.e15	5.1	51
70	Seizures in dominantly inherited Alzheimer disease. <i>Neurology</i> , 2016 , 87, 912-9	6.5	49
69	ABCA7 rare variants and Alzheimer disease risk. <i>Neurology</i> , 2016 , 86, 2134-7	6.5	47
68	PDGFB partial deletion: a new, rare mechanism causing brain calcification with leukoencephalopathy. <i>Journal of Molecular Neuroscience</i> , 2014 , 53, 171-5	3.3	42

67	Juvenile Huntington disease in an 18-month-old boy revealed by global developmental delay and reduced cerebellar volume. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 815-8	2.5	41
66	A de novo nonsense PDGFB mutation causing idiopathic basal ganglia calcification with laryngeal dystonia. <i>European Journal of Human Genetics</i> , 2014 , 22, 1236-8	5.3	40
65	XPR1 mutations are a rare cause of primary familial brain calcification. <i>Journal of Neurology</i> , 2016 , 263, 1559-64	5.5	39
64	SORL1 genetic variants and Alzheimer disease risk: a literature review and meta-analysis of sequencing data. <i>Acta Neuropathologica</i> , 2019 , 138, 173-186	14.3	37
63	Primary brain calcification: an international study reporting novel variants and associated phenotypes. <i>European Journal of Human Genetics</i> , 2018 , 26, 1462-1477	5.3	37
62	Biallelic MYORG mutation carriers exhibit primary brain calcification with a distinct phenotype. <i>Brain</i> , 2019 , 142, 1573-1586	11.2	36
61	hnRNPA2B1 and hnRNPA1 mutations are rare in patients with "multisystem proteinopathy" and frontotemporal lobar degeneration phenotypes. <i>Neurobiology of Aging</i> , 2014 , 35, 934.e5-6	5.6	34
60	Somatic variants in autosomal dominant genes are a rare cause of sporadic Alzheimer's disease. <i>Alzheimers and Dementia</i> , 2018 , 14, 1632-1639	1.2	32
59	From Common to Rare Variants: The Genetic Component of Alzheimer Disease. <i>Human Heredity</i> , 2016 , 81, 129-141	1.1	30
58	The role of de novo mutations in adult-onset neurodegenerative disorders. <i>Acta Neuropathologica</i> , 2019 , 137, 183-207	14.3	29
57	New insights into the genetic etiology of Alzheimer's disease and related dementias.. <i>Nature Genetics</i> , 2022 ,	36.3	27
56	Common variants in Alzheimer's disease and risk stratification by polygenic risk scores. <i>Nature Communications</i> , 2021 , 12, 3417	17.4	23
55	Causative Mutations and Genetic Risk Factors in Sporadic Early Onset Alzheimer's Disease Before 51 Years. <i>Journal of Alzheimers Disease</i> , 2019 , 71, 227-243	4.3	22
54	APP Mutations in Cerebral Amyloid Angiopathy with or without Cortical Calcifications: Report of Three Families and a Literature Review. <i>Journal of Alzheimers Disease</i> , 2017 , 56, 37-46	4.3	20
53	Estimation of minimal disease prevalence from population genomic data: Application to primary familial brain calcification. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2018 , 177, 68-74	3.5	20
52	Overall mutational spectrum of SLC20A2, PDGFB and PDGFRB in idiopathic basal ganglia calcification. <i>Neurogenetics</i> , 2014 , 15, 215-6	3	19
51	Slc20a2, Encoding the Phosphate Transporter PiT2, Is an Important Genetic Determinant of Bone Quality and Strength. <i>Journal of Bone and Mineral Research</i> , 2019 , 34, 1101-1114	6.3	18
50	Identification of partial SLC20A2 deletions in primary brain calcification using whole-exome sequencing. <i>European Journal of Human Genetics</i> , 2016 , 24, 1630-1634	5.3	17

49	Age and the association between apolipoprotein E genotype and Alzheimer disease: A cerebrospinal fluid biomarker-based case-control study. <i>PLoS Medicine</i> , 2020 , 17, e1003289	11.6	15
48	Interplay between primary familial brain calcification-associated SLC20A2 and XPR1 phosphate transporters requires inositol polyphosphates for control of cellular phosphate homeostasis. <i>Journal of Biological Chemistry</i> , 2020 , 295, 9366-9378	5.4	14
47	Brain Calcifications in Adult-Onset Genetic Leukoencephalopathies: A Review. <i>JAMA Neurology</i> , 2017 , 74, 1000-1008	17.2	12
46	Differential Diagnosis of Dementia with High Levels of Cerebrospinal Fluid Tau Protein. <i>Journal of Alzheimer's Disease</i> , 2016 , 51, 905-13	4.3	12
45	Biallelic Loss of Function of SORL1 in an Early Onset Alzheimer's Disease Patient. <i>Journal of Alzheimer's Disease</i> , 2018 , 62, 821-831	4.3	10
44	Detection of copy-number variations from NGS data using read depth information: a diagnostic performance evaluation. <i>European Journal of Human Genetics</i> , 2021 , 29, 99-109	5.3	10
43	Psychosis revealing familial idiopathic basal ganglia calcification. <i>General Hospital Psychiatry</i> , 2013 , 35, 575.e3-5	5.6	9
42	Brain calcifications and variants. <i>Neurology: Genetics</i> , 2017 , 3, e166	3.8	9
41	Microangiopathy in primary familial brain calcification: Evidence from skin biopsies. <i>Neurology: Genetics</i> , 2017 , 3, e134	3.8	8
40	Rare genetic susceptibility variants assessment in autism spectrum disorder: detection rate and practical use. <i>Translational Psychiatry</i> , 2020 , 10, 77	8.6	8
39	Clinical and pathologic features of Aicardi-Goutières syndrome due to an IFIH1 mutation: A pediatric case report. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170A, 1317-24	2.5	8
38	Morphological features in juvenile Huntington disease associated with cerebellar atrophy - magnetic resonance imaging morphometric analysis. <i>Pediatric Radiology</i> , 2018 , 48, 1463-1471	2.8	8
37	Characterization of XPR1/SLC53A1 variants located outside of the SPX domain in patients with primary familial brain calcification. <i>Scientific Reports</i> , 2019 , 9, 6776	4.9	7
36	Dementia in middle-aged patients with schizophrenia. <i>Journal of Alzheimer's Disease</i> , 2014 , 39, 809-22	4.3	7
35	Confirmation and further delineation of the SMG9-deficiency syndrome, a rare and severe developmental disorder. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 2257-2262	2.5	6
34	Early-Onset Cerebral Amyloid Angiopathy and Alzheimer Disease Related to an APP Locus Triplication. <i>Neurology: Genetics</i> , 2021 , 7, e609	3.8	6
33	Reply: New homozygous indel in MYORG linked to brain calcification, thyroidopathy and neuropathy. <i>Brain</i> , 2019 , 142, e52	11.2	5
32	Severe Phenotype in Patients with Large Deletions of. <i>Cancers</i> , 2021 , 13,	6.6	5

31	Exome sequencing identifies three novel AD-associated genes. <i>Alzheimer's and Dementia</i> , 2020 , 16, e041592	4	4
30	Exome sequencing identifies the first genetic determinants of sirenomyelia in humans. <i>Human Mutation</i> , 2020 , 41, 926-933	4.7	4
29	A Simple, Universal, and Cost-Efficient Digital PCR Method for the Targeted Analysis of Copy Number Variations. <i>Clinical Chemistry</i> , 2019 , 65, 1153-1160	5.5	4
28	Genotype-Phenotype Relations in Primary Familial Brain Calcification: Systematic MDSGene Review. <i>Movement Disorders</i> , 2021 , 36, 2468-2480	7	4
27	Significant contribution of intragenic deletions to ARID1B mutation spectrum. <i>Genetics in Medicine</i> , 2019 , 21, 2654-2655	8.1	2
26	Haploinsufficiency of the Primary Familial Brain Calcification Gene SLC20A2 Mediated by Disruption of a Regulatory Element. <i>Movement Disorders</i> , 2020 , 35, 1336-1345	7	2
25	Chorea in an 83-year-old woman: don't forget Huntington's disease. <i>Journal of the American Geriatrics Society</i> , 2012 , 60, 983-4	5.6	2
24	Clinical and neuropathological diversity of tauopathy in MAPT duplication carriers. <i>Acta Neuropathologica</i> , 2021 , 142, 259-278	14.3	2
23	Sporadic Cerebral Amyloid Angiopathy With Cortical Occipital Calcifications in the Elderly. <i>Alzheimer Disease and Associated Disorders</i> , 2018 , 32, 83-84	2.5	2
22	A response to the letter by Oliveira et al. <i>General Hospital Psychiatry</i> , 2013 , 35, 212	5.6	1
21	A postzygotic de novo NCDN mutation identified in a sporadic FTLD patient results in neurochondrin haploinsufficiency and altered FUS granule dynamics.. <i>Acta Neuropathologica Communications</i> , 2022 , 10, 20	7.3	1
20	MYT1L-associated neurodevelopmental disorder: description of 40 new cases and literature review of clinical and molecular aspects. <i>Human Genetics</i> , 2021 , 1	6.3	1
19	Exome sequencing identifies rare damaging variants in the ATP8B4 and ABCA1 genes as novel risk factors for Alzheimer's Disease		1
18	Hypersociability associated with developmental delay, macrocephaly and facial dysmorphism points to CHD3 mutations. <i>European Journal of Medical Genetics</i> , 2021 , 64, 104166	2.6	1
17	Impaired SorLA maturation and trafficking as a new mechanism for SORL1 missense variants in Alzheimer disease		1
16	Penetrance estimation of SORL1 loss-of-function variants using a family-based strategy adjusted on APOE genotypes suggest a non-monogenic inheritance		1
15	Distinct functional classes of PDGFRB pathogenic variants in primary familial brain calcification. <i>Human Molecular Genetics</i> , 2021 ,	5.6	1
14	Impaired SorLA maturation and trafficking as a new mechanism for SORL1 missense variants in Alzheimer disease.. <i>Acta Neuropathologica Communications</i> , 2021 , 9, 196	7.3	0

13	Recurrence of an early postzygotic rescue of an inherited unbalanced translocation resulting in mosaic segmental uniparental isodisomy of chromosome 11q in siblings. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 3057-3061	2.5	0
12	NGLY1 Deficiency: A Rare Newly Described Condition with a Typical Presentation. <i>Life</i> , 2021 , 11,	3	0
11	Assessment of SORL1 rare variants segregation in Alzheimer disease families and in vitro models suggests diverse penetrance and oligogenic inheritance. <i>Alzheimer's and Dementia</i> , 2020 , 16, e044561	1.2	
10	A diagnosis of idiopathic basal ganglia calcification in an 82-year-old man. <i>Journal of the American Geriatrics Society</i> , 2013 , 61, 2057-9	5.6	
9	Reply: Primary brain calcification due to a homozygous MYORG mutation causing isolated paroxysmal kinesigenic dyskinesia. <i>Brain</i> , 2020 , 143, e37	11.2	
8	Generation of 17q21.31 duplication iPSC-derived neurons as a model for primary tauopathies.. <i>Stem Cell Research</i> , 2022 , 61, 102762	1.6	
7	Age and the association between apolipoprotein E genotype and Alzheimer disease: A cerebrospinal fluid biomarker-based case-control study 2020 , 17, e1003289		
6	Age and the association between apolipoprotein E genotype and Alzheimer disease: A cerebrospinal fluid biomarker-based case-control study 2020 , 17, e1003289		
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