Gal Nicolas

List of Publications by Year in Descending Order

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	A postzygotic de novo NCDN mutation identified in a sporadic FTLD patient results in neurochondrin haploinsufficiency and altered FUS granule dynamics.. *Acta Neuropathologica Communications*, **2022**, 10, 20	7.3	1
83	New insights into the genetic etiology of Alzheimer's disease and related dementias.. *Nature Genetics*, **2022**,	36.3	27
82	Generation of 17q21.31 duplication iPSC-derived neurons as a model for primary tauopathies.. *Stem Cell Research*, **2022**, 61, 102762	1.6	
81	Impaired SorLA maturation and trafficking as a new mechanism for SORL1 missense variants in Alzheimer disease.. *Acta Neuropathologica Communications*, **2021**, 9, 196	7.3	0
80	MYT1L-associated neurodevelopmental disorder: description of 40 new cases and literature review of clinical and molecular aspects. *Human Genetics*, **2021**, 1	6.3	1
79	Hypersociability associated with developmental delay, macrocephaly and facial dysmorphism points to CHD3 mutations. *European Journal of Medical Genetics*, **2021**, 64, 104166	2.6	1
78	Recurrence of an early postzygotic rescue of an inherited unbalanced translocation resulting in mosaic segmental uniparental isodisomy of chromosome 11q in siblings. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3057-3061	2.5	0
77	Clinical and neuropathological diversity of tauopathy in MAPT duplication carriers. *Acta Neuropathologica*, **2021**, 142, 259-278	14.3	2
76	Common variants in Alzheimer's disease and risk stratification by polygenic risk scores. *Nature Communications*, **2021**, 12, 3417	17.4	23
75	Severe Phenotype in Patients with Large Deletions of. *Cancers*, **2021**, 13,	6.6	5
74	Detection of copy-number variations from NGS data using read depth information: a diagnostic performance evaluation. *European Journal of Human Genetics*, **2021**, 29, 99-109	5.3	10
73	NGLY1 Deficiency: A Rare Newly Described Condition with a Typical Presentation. *Life*, **2021**, 11,	3	0
72	Genotype-Phenotype Relations in Primary Familial Brain Calcification: Systematic MDSGene Review. *Movement Disorders*, **2021**, 36, 2468-2480	7	4
71	Early-Onset Cerebral Amyloid Angiopathy and Alzheimer Disease Related to an APP Locus Triplication. *Neurology: Genetics*, **2021**, 7, e609	3.8	6
70	Distinct functional classes of PDGFRB pathogenic variants in primary familial brain calcification. *Human Molecular Genetics*, **2021**,	5.6	1
69	Exome sequencing identifies three novel AD-associated genes. *Alzheimers and Dementia*, **2020**, 16, e041592	1.2	4
68	Assessment of SORL1 rare variants segregation in Alzheimer disease families and in vitro models suggests diverse penetrance and oligogenic inheritance. *Alzheimers and Dementia*, **2020**, 16, e044561	1.2	

67	Interplay between primary familial brain calcification-associated SLC20A2 and XPR1 phosphate transporters requires inositol polyphosphates for control of cellular phosphate homeostasis. *Journal of Biological Chemistry*, **2020**, 295, 9366-9378	5.4	14
66	Haploinsufficiency of the Primary Familial Brain Calcification Gene SLC20A2 Mediated by Disruption of a Regulatory Element. *Movement Disorders*, **2020**, 35, 1336-1345	7	2
65	Rare genetic susceptibility variants assessment in autism spectrum disorder: detection rate and practical use. *Translational Psychiatry*, **2020**, 10, 77	8.6	8
64	Exome sequencing identifies the first genetic determinants of sirenomelia in humans. *Human Mutation*, **2020**, 41, 926-933	4.7	4
63	Age and the association between apolipoprotein E genotype and Alzheimer disease: A cerebrospinal fluid biomarker-based case-control study. *PLoS Medicine*, **2020**, 17, e1003289	11.6	15
62	Whole exome sequencing study identifies novel rare and common Alzheimer's-Associated variants involved in immune response and transcriptional regulation. *Molecular Psychiatry*, **2020**, 25, 1859-1875	15.1	106
61	Reply: Primary brain calcification due to a homozygous MYORG mutation causing isolated paroxysmal kinesigenic dyskinesia. *Brain*, **2020**, 143, e37	11.2	
60	Age and the association between apolipoprotein E genotype and Alzheimer disease: A cerebrospinal fluid biomarkerBased caseControl study **2020**, 17, e1003289		
59	Age and the association between apolipoprotein E genotype and Alzheimer disease: A cerebrospinal fluid biomarkerBased caseControl study **2020**, 17, e1003289		
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55	Age and the association between apolipoprotein E genotype and Alzheimer disease: A cerebrospinal fluid biomarkerBased caseControl study **2020**, 17, e1003289		
54	Age and the association between apolipoprotein E genotype and Alzheimer disease: A cerebrospinal fluid biomarkerBased caseControl study **2020**, 17, e1003289		
53	Significant contribution of intragenic deletions to ARID1B mutation spectrum. *Genetics in Medicine*, **2019**, 21, 2654-2655	8.1	2
52	Characterization of XPR1/SLC53A1 variants located outside of the SPX domain in patients with primary familial brain calcification. *Scientific Reports*, **2019**, 9, 6776	4.9	7
51	Biallelic MYORG mutation carriers exhibit primary brain calcification with a distinct phenotype. *Brain*, **2019**, 142, 1573-1586	11.2	36
50	SORL1 genetic variants and Alzheimer disease risk: a literature review and meta-analysis of sequencing data. *Acta Neuropathologica*, **2019**, 138, 173-186	14.3	37

49	Slc20a2, Encoding the Phosphate Transporter PiT2, Is an Important Genetic Determinant of Bone Quality and Strength. *Journal of Bone and Mineral Research*, **2019**, 34, 1101-1114	6.3	18
48	Causative Mutations and Genetic Risk Factors in Sporadic Early Onset Alzheimer's Disease Before 51 Years. *Journal of Alzheimer's Disease*, **2019**, 71, 227-243	4.3	22
47	Confirmation and further delineation of the SMG9-deficiency syndrome, a rare and severe developmental disorder. *American Journal of Medical Genetics, Part A*, **2019**, 179, 2257-2262	2.5	6
46	A Simple, Universal, and Cost-Efficient Digital PCR Method for the Targeted Analysis of Copy Number Variations. *Clinical Chemistry*, **2019**, 65, 1153-1160	5.5	4
45	Reply: New homozygous indel in MYORG linked to brain calcification, thyroidopathy and neuropathy. *Brain*, **2019**, 142, e52	11.2	5
44	Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates A tau, immunity and lipid processing. *Nature Genetics*, **2019**, 51, 414-430	36.3	917
43	The role of de novo mutations in adult-onset neurodegenerative disorders. *Acta Neuropathologica*, **2019**, 137, 183-207	14.3	29
42	Biallelic Loss of Function of SORL1 in an Early Onset Alzheimer's Disease Patient. *Journal of Alzheimer's Disease*, **2018**, 62, 821-831	4.3	10
41	Primary brain calcification: an international study reporting novel variants and associated phenotypes. *European Journal of Human Genetics*, **2018**, 26, 1462-1477	5.3	37
40	Morphological features in juvenile Huntington disease associated with cerebellar atrophy - magnetic resonance imaging morphometric analysis. *Pediatric Radiology*, **2018**, 48, 1463-1471	2.8	8
39	Somatic variants in autosomal dominant genes are a rare cause of sporadic Alzheimer's disease. *Alzheimer's and Dementia*, **2018**, 14, 1632-1639	1.2	32
38	Estimation of minimal disease prevalence from population genomic data: Application to primary familial brain calcification. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2018**, 177, 68-74	3.5	20
37	Sporadic Cerebral Amyloid Angiopathy With Cortical Occipital Calcifications in the Elderly. *Alzheimer Disease and Associated Disorders*, **2018**, 32, 83-84	2.5	2
36	Microangiopathy in primary familial brain calcification: Evidence from skin biopsies. *Neurology: Genetics*, **2017**, 3, e134	3.8	8
35	APP Mutations in Cerebral Amyloid Angiopathy with or without Cortical Calcifications: Report of Three Families and a Literature Review. *Journal of Alzheimer's Disease*, **2017**, 56, 37-46	4.3	20
34	Contribution to Alzheimer's disease risk of rare variants in TREM2, SORL1, and ABCA7 in 1779 cases and 1273 controls. *Neurobiology of Aging*, **2017**, 59, 220.e1-220.e9	5.6	83
33	Brain Calcifications in Adult-Onset Genetic Leukoencephalopathies: A Review. *JAMA Neurology*, **2017**, 74, 1000-1008	17.2	12
32	Brain calcifications and variants. *Neurology: Genetics*, **2017**, 3, e166	3.8	9

31	APP, PSEN1, and PSEN2 mutations in early-onset Alzheimer disease: A genetic screening study of familial and sporadic cases. *PLoS Medicine*, **2017**, 14, e1002270	11.6	204
30	Screening of dementia genes by whole-exome sequencing in early-onset Alzheimer disease: input and lessons. *European Journal of Human Genetics*, **2016**, 24, 710-6	5.3	61
29	SORL1 rare variants: a major risk factor for familial early-onset Alzheimer's disease. *Molecular Psychiatry*, **2016**, 21, 831-6	15.1	71
28	Clinical and pathologic features of Aicardi-Goutières syndrome due to an IFIH1 mutation: A pediatric case report. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 1317-24	2.5	8
27	Identification of partial SLC20A2 deletions in primary brain calcification using whole-exome sequencing. *European Journal of Human Genetics*, **2016**, 24, 1630-1634	5.3	17
26	Differential Diagnosis of Dementia with High Levels of Cerebrospinal Fluid Tau Protein. *Journal of Alzheimer's Disease*, **2016**, 51, 905-13	4.3	12
25	From Common to Rare Variants: The Genetic Component of Alzheimer Disease. *Human Heredity*, **2016**, 81, 129-141	1.1	30
24	XPR1 mutations are a rare cause of primary familial brain calcification. *Journal of Neurology*, **2016**, 263, 1559-64	5.5	39
23	ABCA7 rare variants and Alzheimer disease risk. *Neurology*, **2016**, 86, 2134-7	6.5	47
22	TYROBP genetic variants in early-onset Alzheimer's disease. *Neurobiology of Aging*, **2016**, 48, 222.e9-222.e15	5.1	51
21	Seizures in dominantly inherited Alzheimer disease. *Neurology*, **2016**, 87, 912-9	6.5	49
20	Update and Mutational Analysis of SLC20A2: A Major Cause of Primary Familial Brain Calcification. *Human Mutation*, **2015**, 36, 489-95	4.7	54
19	Mutations in XPR1 cause primary familial brain calcification associated with altered phosphate export. *Nature Genetics*, **2015**, 47, 579-81	36.3	176
18	Brain calcification process and phenotypes according to age and sex: Lessons from SLC20A2, PDGFB, and PDGFRB mutation carriers. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2015**, 168, 586-94	3.5	61
17	Dementia in middle-aged patients with schizophrenia. *Journal of Alzheimer's Disease*, **2014**, 39, 809-22	4.3	7
16	PDGFB partial deletion: a new, rare mechanism causing brain calcification with leukoencephalopathy. *Journal of Molecular Neuroscience*, **2014**, 53, 171-5	3.3	42
15	Overall mutational spectrum of SLC20A2, PDGFB and PDGFRB in idiopathic basal ganglia calcification. *Neurogenetics*, **2014**, 15, 215-6	3	19
14	hnRNPA2B1 and hnRNPA1 mutations are rare in patients with "multisystem proteinopathy" and frontotemporal lobar degeneration phenotypes. *Neurobiology of Aging*, **2014**, 35, 934.e5-6	5.6	34

13	A de novo nonsense PDGFB mutation causing idiopathic basal ganglia calcification with laryngeal dystonia. *European Journal of Human Genetics*, **2014**, 22, 1236-8	5.3	40
12	Mutations in the gene encoding PDGF-B cause brain calcifications in humans and mice. *Nature Genetics*, **2013**, 45, 1077-82	36.3	214
11	Mutation of the PDGFRB gene as a cause of idiopathic basal ganglia calcification. *Neurology*, **2013**, 80, 181-7	6.5	192
10	A response to the letter by Oliveira et al. *General Hospital Psychiatry*, **2013**, 35, 212	5.6	1
9	Psychosis revealing familial idiopathic basal ganglia calcification. *General Hospital Psychiatry*, **2013**, 35, 575.e3-5	5.6	9
8	A diagnosis of idiopathic basal ganglia calcification in an 82-year-old man. *Journal of the American Geriatrics Society*, **2013**, 61, 2057-9	5.6	
7	Phenotypic spectrum of probable and genetically-confirmed idiopathic basal ganglia calcification. *Brain*, **2013**, 136, 3395-407	11.2	146
6	SQSTM1 mutations in French patients with frontotemporal dementia or frontotemporal dementia with amyotrophic lateral sclerosis. *JAMA Neurology*, **2013**, 70, 1403-10	17.2	131
5	Chorea in an 83-year-old woman: don't forget Huntington's disease. *Journal of the American Geriatrics Society*, **2012**, 60, 983-4	5.6	2
4	Juvenile Huntington disease in an 18-month-old boy revealed by global developmental delay and reduced cerebellar volume. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 815-8	2.5	41
3	Exome sequencing identifies rare damaging variants in the ATP8B4 and ABCA1 genes as novel risk factors for Alzheimer Disease		1
2	Impaired SorLA maturation and trafficking as a new mechanism for SORL1 missense variants in Alzheimer disease		1
1	Penetrance estimation of SORL1 loss-of-function variants using a family-based strategy adjusted on APOE genotypes suggest a non-monogenic inheritance		1