

Cyril Pottier

List of Publications by Year in descending order

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Version: 2024-02-01

30
papers

2,703
citations

331670

21
h-index

454955

30
g-index

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docs citations

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times ranked

4905
citing authors

#	ARTICLE	IF	CITATIONS
1	TIA1 Mutations in Amyotrophic Lateral Sclerosis and Frontotemporal Dementia Promote Phase Separation and Alter Stress Granule Dynamics. <i>Neuron</i> , 2017, 95, 808-816.e9.	8.1	493
2	Whole-genome sequencing reveals important role for TBK1 and OPTN mutations in frontotemporal lobar degeneration without motor neuron disease. <i>Acta Neuropathologica</i> , 2015, 130, 77-92.	7.7	267
3	High frequency of potentially pathogenic SORL1 mutations in autosomal dominant early-onset Alzheimer disease. <i>Molecular Psychiatry</i> , 2012, 17, 875-879.	7.9	253
4	Mutation of the <i>PDGFRB</i> gene as a cause of idiopathic basal ganglia calcification. <i>Neurology</i> , 2013, 80, 181-187.	1.1	239
5	Phenotypic spectrum of probable and genetically-confirmed idiopathic basal ganglia calcification. <i>Brain</i> , 2013, 136, 3395-3407.	7.6	183
6	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. <i>Nature Communications</i> , 2016, 7, 11253.	12.8	174
7	TREM2 R47H Variant as a Risk Factor for Early-Onset Alzheimer's Disease. <i>Journal of Alzheimer's Disease</i> , 2013, 35, 45-49.	2.6	136
8	Genetics of <i>FTLD</i> : overview and what else we can expect from genetic studies. <i>Journal of Neurochemistry</i> , 2016, 138, 32-53.	3.9	118
9	Potential genetic modifiers of disease risk and age at onset in patients with frontotemporal lobar degeneration and GRN mutations: a genome-wide association study. <i>Lancet Neurology</i> , The, 2018, 17, 548-558.	10.2	97
10	Genome-wide analyses as part of the international <i>FTLD-TDP</i> whole-genome sequencing consortium reveals novel disease risk factors and increases support for immune dysfunction in <i>FTLD</i> . <i>Acta Neuropathologica</i> , 2019, 137, 879-899.	7.7	90
11	De novo deleterious genetic variations target a biological network centered on $A\beta$ peptide in early-onset Alzheimer disease. <i>Molecular Psychiatry</i> , 2015, 20, 1046-1056.	7.9	89
12	TYROBP genetic variants in early-onset Alzheimer's disease. <i>Neurobiology of Aging</i> , 2016, 48, 222.e9-222.e15.	3.1	69
13	Prosaposin is a regulator of progranulin levels and oligomerization. <i>Nature Communications</i> , 2016, 7, 11992.	12.8	68
14	PDGFB Partial Deletion: a New, Rare Mechanism Causing Brain Calcification with Leukoencephalopathy. <i>Journal of Molecular Neuroscience</i> , 2014, 53, 171-175.	2.3	50
15	Alzheimer disease: modeling an $A\beta$ -centered biological network. <i>Molecular Psychiatry</i> , 2016, 21, 861-871.	7.9	47
16	A de novo nonsense PDGFB mutation causing idiopathic basal ganglia calcification with laryngeal dystonia. <i>European Journal of Human Genetics</i> , 2014, 22, 1236-1238.	2.8	46
17	Extensive transcriptomic study emphasizes importance of vesicular transport in C9orf72 expansion carriers. <i>Acta Neuropathologica Communications</i> , 2019, 7, 150.	5.2	40
18	Clinical and neuropathological features of ALS/FTD with TIA1 mutations. <i>Acta Neuropathologica Communications</i> , 2017, 5, 96.	5.2	38

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19	Mutation in the 3' untranslated region of APP as a genetic determinant of cerebral amyloid angiopathy. <i>European Journal of Human Genetics</i> , 2016, 24, 92-98.	2.8	26
20	Clinicopathologic correlations in a family with a <i>TBK1</i> mutation presenting as primary progressive aphasia and primary lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2019, 20, 568-575.	1.7	24
21	Amyloid- β Protein Precursor Gene Expression in Alzheimer's Disease and Other Conditions. <i>Journal of Alzheimer's Disease</i> , 2012, 28, 561-566.	2.6	22
22	Overall mutational spectrum of SLC20A2, PDGFB and PDGFRB in idiopathic basal ganglia calcification. <i>Neurogenetics</i> , 2014, 15, 215-216.	1.4	22
23	Three VCP Mutations in Patients with Frontotemporal Dementia. <i>Journal of Alzheimer's Disease</i> , 2018, 65, 1139-1146.	2.6	19
24	Underlying genetic variation in familial frontotemporal dementia: sequencing of 198 patients. <i>Neurobiology of Aging</i> , 2021, 97, 148.e9-148.e16.	3.1	17
25	Identification of compound heterozygous variants in <i>OPTN</i> in an ALS-FTD patient from the CReATe consortium: a case report. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2018, 19, 469-471.	1.7	15
26	Identification of missing variants by combining multiple analytic pipelines. <i>BMC Bioinformatics</i> , 2018, 19, 139.	2.6	10
27	<i>PRKAR1B</i> mutations are a rare cause of FUS negative neuronal intermediate filament inclusion disease. <i>Brain</i> , 2015, 138, e357-e357.	7.6	9
28	Shared brain transcriptomic signature in TDP-43 type A FTLD patients with or without <i>GRN</i> mutations. <i>Brain</i> , 2022, 145, 2472-2485.	7.6	6
29	Impact of variant-level batch effects on identification of genetic risk factors in large sequencing studies. <i>PLoS ONE</i> , 2021, 16, e0249305.	2.5	5
30	The presenilin 1 p.Gly206Ala mutation is a frequent cause of early-onset Alzheimer's disease in Hispanics in Florida. <i>American Journal of Neurodegenerative Disease</i> , 2016, 5, 94-101.	0.1	4