Kin Y Mok

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/8687041/publications.pdf

Version: 2024-02-01

79 papers	12,086 citations	81900 39 h-index	71685 76 g-index

87 all docs	87 docs citations	87 times ranked	17451 citing authors

#	Article	IF	Citations
1	A Hexanucleotide Repeat Expansion in C9ORF72 Is the Cause of Chromosome 9p21-Linked ALS-FTD. Neuron, 2011, 72, 257-268.	8.1	3,833
2	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
3	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	12.6	1,085
4	Frequency of the C9orf72 hexanucleotide repeat expansion in patients with amyotrophic lateral sclerosis and frontotemporal dementia: a cross-sectional study. Lancet Neurology, The, 2012, 11, 323-330.	10.2	1,039
5	A robust model for read count data in exome sequencing experiments and implications for copy number variant calling. Bioinformatics, 2012, 28, 2747-2754.	4.1	534
6	Frontotemporal dementia with the C9ORF72 hexanucleotide repeat expansion: clinical, neuroanatomical and neuropathological features. Brain, 2012, 135, 736-750.	7.6	392
7	Clinico-pathological features in amyotrophic lateral sclerosis with expansions in C9ORF72. Brain, 2012, 135, 751-764.	7.6	293
8	Chromosome 9p21 in sporadic amyotrophic lateral sclerosis in the UK and seven other countries: a genome-wide association study. Lancet Neurology, The, 2010, 9, 986-994.	10.2	205
9	Identification of Risk Loci for Parkinson Disease in Asians and Comparison of Risk Between Asians and Europeans. JAMA Neurology, 2020, 77, 746.	9.0	170
10	Parkinsonâ€™s disease in GTP cyclohydrolase 1 mutation carriers. Brain, 2014, 137, 2480-2492.	7.6	169
11	The importance of understanding individual differences in Down syndrome. F1000Research, 2016, 5, 389.	1.6	151
12	A genome-wide association study in multiple system atrophy. Neurology, 2016, 87, 1591-1598.	1.1	139
13	The chromosome 9 ALS and FTD locus is probably derived from a single founder. Neurobiology of Aging, 2012, 33, 209.e3-209.e8.	3.1	115
14	Association of Dementia With Mortality Among Adults With Down Syndrome Older Than 35 Years. JAMA Neurology, 2019, 76, 152.	9.0	110
15	NeuroChip, an updated version of the NeuroX genotyping platform to rapidly screen for variants associated with neurological diseases. Neurobiology of Aging, 2017, 57, 247.e9-247.e13.	3.1	108
16	Astrogliopathy predominates the earliest stage of corticobasal degeneration pathology. Brain, 2016, 139, 3237-3252.	7.6	107
17	Autonomic dysfunction in ALS: A preliminary study on the effects of intrathecal BDNF. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2005, 6, 100-103.	2.1	102
18	Mitochondria function associated genes contribute to Parkinsonâ€™s Disease risk and later age at onset. Npj Parkinson's Disease, 2019, 5, 8.	5.3	95

#	Article	IF	Citations
19	Non-coding variability at the APOE locus contributes to the Alzheimerâ€™s risk. Nature Communications, 2019, 10, 3310.	12.8	91
20	LRBA gene deletion in a patient presenting with autoimmunity without hypogammaglobulinemia. Journal of Allergy and Clinical Immunology, 2012, 130, 1428-1432.	2.9	90
21	Health comorbidities and cognitive abilities across the lifespan in Down syndrome. Journal of Neurodevelopmental Disorders, 2020, 12, 4.	3.1	85
22	Moving beyond neurons: the role of cell type-specific gene regulation in Parkinsonâ€™s disease heritability. Npj Parkinson's Disease, 2019, 5, 6.	5.3	83
23	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
24	Identification of genetic risk factors in the Chinese population implicates a role of immune system in Alzheimerâ€™s disease pathogenesis. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 1697-1706.	7.1	71
25	Cognitive markers of preclinical and prodromal Alzheimer's disease in Down syndrome. Alzheimer's and Dementia, 2019, 15, 245-257.	0.8	68
26	Frequency of Loss of Function Variants in LRRK2 in Parkinson Disease. JAMA Neurology, 2018, 75, 1416.	9.0	66
27	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
28	Largeâ€scale plasma proteomic profiling identifies a highâ€performance biomarker panel for Alzheimer's disease screening and staging. Alzheimer's and Dementia, 2022, 18, 88-102.	0.8	65
29	Aging related cognitive changes associated with Alzheimer's disease in Down syndrome. Annals of Clinical and Translational Neurology, 2018, 5, 741-751.	3.7	64
30	Patient-specific Alzheimer-like pathology in trisomy 21 cerebral organoids reveals BACE2 as a gene dose-sensitive AD suppressor in human brain. Molecular Psychiatry, 2021, 26, 5766-5788.	7.9	63
31	Genetic determinants of survival in progressive supranuclear palsy: a genome-wide association study. Lancet Neurology, The, 2021, 20, 107-116.	10.2	62
32	Plasma biomarkers for amyloid, tau, and cytokines in Down syndrome and sporadic Alzheimerâ€™s disease. Alzheimer's Research and Therapy, 2019, 11, 26.	6.2	56
33	C9orf72 intermediate repeats are associated with corticobasal degeneration, increased C9orf72 expression and disruption of autophagy. Acta Neuropathologica, 2019, 138, 795-811.	7.7	50
34	Dystonia genes functionally converge in specific neurons and share neurobiology with psychiatric disorders. Brain, 2020, 143, 2771-2787.	7.6	50
35	Screening a UK amyotrophic lateral sclerosis cohort provides evidence of multiple origins of the C9orf72 expansion. Neurobiology of Aging, 2015, 36, 546.e1-546.e7.	3.1	48
36	Additional rare variant analysis in Parkinsonâ€™s disease cases with and without known pathogenic mutations: evidence for oligogenic inheritance. Human Molecular Genetics, 2016, 25, ddw348.	2.9	48

#	Article	IF	Citations
37	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
38	Screening for C9ORF72 repeat expansion in FTLD. Neurobiology of Aging, 2012, 33, 1850.e1-1850.e11.	3.1	46
39	High frequency of the expanded C9ORF72 hexanucleotide repeat in familial and sporadic Greek ALS patients. Neurobiology of Aging, 2012, 33, 1851.e1-1851.e5.	3.1	44
40	Finding genetically-supported drug targets for Parkinsonâ€™s disease using Mendelian randomization of the druggable genome. Nature Communications, 2021, 12, 7342.	12.8	44
41	Genomewide association study in cervical dystonia demonstrates possible association with sodium leak channel. Movement Disorders, 2014, 29, 245-251.	3.9	43
42	Neurofilament light as a blood biomarker for neurodegeneration in Down syndrome. Alzheimer's Research and Therapy, 2018, 10, 39.	6.2	43
43	Polymorphisms in BACE2 may affect the age of onset Alzheimer's dementia in Down syndrome. Neurobiology of Aging, 2014, 35, 1513.e1-1513.e5.	3.1	41
44	Identification of sixteen novel candidate genes for late onset Parkinsonâ€™s disease. Molecular Neurodegeneration, 2021, 16, 35.	10.8	41
45	Variation at the TRIM11 locus modifies progressive supranuclear palsy phenotype. Annals of Neurology, 2018, 84, 485-496.	5.3	37
46	Evidence that PICALM affects age at onset of Alzheimer's dementia in Down syndrome. Neurobiology of Aging, 2013, 34, 2441.e1-2441.e5.	3.1	35
47	Typical features of Parkinson disease and diagnostic challenges with microdeletion 22q11.2. Neurology, 2018, 90, e2059-e2067.	1.1	35
48	Identification of a novel THAP1 mutation at R29 aminoâ€acid residue in sporadic patients with earlyâ€onset dystonia. Movement Disorders, 2009, 24, 2428-2429.	3.9	32
49	Insufficient evidence for pathogenicity of SNCA His50Gln (H50Q) in Parkinson's disease. Neurobiology of Aging, 2018, 64, 159.e5-159.e8.	3.1	30
50	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. Annals of Neurology, 2021, 90, 35-42.	5.3	29
51	Cerebral Venous Thrombosis in Hong Kong. Cerebrovascular Diseases, 2001, 11, 282-283.	1.7	26
52	Lack of evidence for a role of genetic variation in TMEM230 in the risk for Parkinson's disease in the Caucasian population. Neurobiology of Aging, 2017, 50, 167.e11-167.e13.	3.1	24
53	Regulatory sites for splicing in human basal ganglia are enriched for disease-relevant information. Nature Communications, 2020, 11, 1041.	12.8	22
54	SNCA and mTOR Pathway Single Nucleotide Polymorphisms Interact to Modulate the Age at Onset of Parkinson's Disease. Movement Disorders, 2019, 34, 1333-1344.	3.9	21

#	Article	IF	Citations
55	LRP10 in Î±-synucleinopathies. Lancet Neurology, The, 2018, 17, 1032.	10.2	15
56	Fulminant corticobasal degeneration: a distinct variant with predominant neuronal tau aggregates. Acta Neuropathologica, 2020, 139, 717-734.	7.7	15
57	Familial Lund frontotemporal dementia caused by C9ORF72 hexanucleotide expansion. Neurobiology of Aging, 2012, 33, 1850.e13-1850.e16.	3.1	14
58	HLA-DRB*1501 associations with magnetic resonance imaging measures of grey matter pathology in multiple sclerosis. Multiple Sclerosis and Related Disorders, 2016, 7, 47-52.	2.0	14
59	Genetic and polygenic risk score analysis for Alzheimer's disease in the Chinese population. Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring, 2020, 12, e12074.	2.4	14
60	A multi-level developmental approach to exploring individual differences in Down syndrome: genes, brain, behaviour, and environment. Research in Developmental Disabilities, 2020, 104, 103638.	2.2	13
61	Madras motor neuron disease (MMND) is distinct from the riboflavin transporter genetic defects that cause Brownâ€“Vialettoâ€“Van Laere syndrome. Journal of the Neurological Sciences, 2013, 334, 119-122.	0.6	12
62	Homozygosity analysis in amyotrophic lateral sclerosis. European Journal of Human Genetics, 2013, 21, 1429-1435.	2.8	12
63	Nocardia Peritonitis Complicating Continuous Ambulatory Peritoneal Dialysis. Peritoneal Dialysis International, 1990, 10, 99-99.	2.3	11
64	Parkinson's disease without nigral degeneration: a pathological correlate of scans without evidence of dopaminergic deficit (SWEDD)?. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 633-641.	1.9	11
65	Differential Associations of Apolipoprotein E Îµ4 Genotype With Attentional Abilities Across the Life Span of Individuals With Down Syndrome. JAMA Network Open, 2020, 3, e2018221.	5.9	7
66	A multimodal neurophysiological assessment in terminal renal failure. Acta Neurologica Scandinavica, 1991, 83, 89-95.	2.1	6
67	Behavioral Variant Frontotemporal Lobar Degeneration with Amyotrophic Lateral Sclerosis with a Chromosome 9p21 Hexanucleotide Repeat. Frontiers in Neurology, 2012, 3, 136.	2.4	6
68	Fibrillation and molecular characteristics are coherent with clinical and pathological features of 4-repeat tauopathy caused by MAPT variant G273R. Neurobiology of Disease, 2020, 146, 105079.	4.4	4
69	A highâ€performance biomarker panel for Alzheimerâ€™s disease screening and staging identified by largeâ€scale plasma proteomic profiling. Alzheimer's and Dementia, 2021, 17, .	0.8	4
70	The East Asian Parkinson Disease Genomics Consortium. Lancet Neurology, The, 2021, 20, 982.	10.2	3
71	Demographics and Medication Use of Patients with Late-Onset Alzheimerâ€™s Disease in Hong Kong. Journal of Alzheimer's Disease, 2022, 87, 1205-1213.	2.6	3
72	Response to the commentary of Yates RL and DeLuca GC on the study: HLA-DRB1*1501 associations with magnetic resonance imaging measures of grey matter pathology in multiple sclerosis. Multiple Sclerosis and Related Disorders, 2018, 19, 168-170.	2.0	2

#	Article	IF	Citations
73	A familial frontotemporal dementia associated with C9orf72 repeat expansion and dysplastic gangliocytoma. Neurobiology of Aging, 2014, 35, 444.e11-444.e14.	3.1	1
74	Blood transcriptome analysis for Alzheimer' disease in Hong Kong Chinese population.. Alzheimer's and Dementia, 2021, 17 Suppl 3, e056643.	0.8	1
75	O1â€05â€01: Frontotemporal dementia with the C9ORF72 hexanucleotide repeat expansion: Clinical, neuroanatomical and neuropathological features. Alzheimer's and Dementia, 2012, 8, P92.	0.8	0
76	A Man with Difficulty Chewing Gum and an Ominous Family History. , 2017, , 103-106.		0
77	Reply to â€œDown Syndrome Cognitive Marker's Significance in Alzheimer's Disease and Dementia Managementâ€. Alzheimer's and Dementia, 2019, 15, 1238-1239.	0.8	0
78	Evaluation of genetic risk for Alzheimerâ€™s disease in the Hong Kong Chinese population. Alzheimer's and Dementia, 2020, 16, e045142.	0.8	0
79	Deep learning for polygenic score analysis for Alzheimer's disease risk prediction in the Chinese population.. Alzheimer's and Dementia, 2021, 17 Suppl 3, e056625.	0.8	0