Camille Charbonnier

List of Publications by Year
in descending order

17	1,585	623734 14	888059 17
papers	citations	h-index	g-index

18	18	18	3534
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Penetrance estimation of Alzheimer disease in SORL1 loss-of-function variant carriers using a family-based strategy and stratification by APOE genotypes. Genome Medicine, 2022, 14, .	8.2	7
2	Whole exome sequencing study identifies novel rare and common Alzheimerâ€™s-Associated variants involved in immune response and transcriptional regulation. Molecular Psychiatry, 2020, 25, 1859-1875.	7.9	191
3	Haploinsufficiency of the Primary Familial Brain Calcification Gene <scp>SLC20A2</scp> Mediated by Disruption of a Regulatory Element. Movement Disorders, 2020, 35, 1336-1345.	3.9	9
4	Biallelic MYORG mutation carriers exhibit primary brain calcification with a distinct phenotype. Brain, 2019, 142, 1573-1586.	7.6	49
5	Copy Number Variants in miR-138 as a Potential Risk Factor for Early-Onset Alzheimerâ€™s Disease. Journal of Alzheimer's Disease, 2019, 68, 1243-1255.	2.6	19
6	SORL1 genetic variants and Alzheimer disease risk: a literature review and meta-analysis of sequencing data. Acta Neuropathologica, 2019, 138, 173-186.	7.7	77
7	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.	1.7	33
8	Contribution of genotoxic anticancer treatments to the development of multiple primary tumours in the context of germline TP53 mutations. European Journal of Cancer, 2018, 101, 254-262.	2.8	32
9	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.	3.1	116
10	From Common to Rare Variants: The Genetic Component of Alzheimer Disease. Human Heredity, 2016, 81, 129-141.	0.8	37
11	ABCA7 rare variants and Alzheimer disease risk. Neurology, 2016, 86, 2134-2137.	1.1	63
12	Seizures in dominantly inherited Alzheimer disease. Neurology, 2016, 87, 912-919.	1.1	81
13	Screening of dementia genes by whole-exome sequencing in early-onset Alzheimer disease: input and lessons. European Journal of Human Genetics, 2016, 24, 710-716.	2.8	77
14	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, 586-594.	1.7	74
15	Revisiting Li-Fraumeni Syndrome From TP53 Mutation Carriers. Journal of Clinical Oncology, 2015, 33, 2345-2352.	1.6	525
16	Germline Mutations of Inhibins in Earlyâ€Onset Ovarian Epithelial Tumors. Human Mutation, 2014, 35, 294-297.	2.5	11
17	Phenotypic spectrum of probable and genetically-confirmed idiopathic basal ganglia calcification. Brain, 2013, 136, 3395-3407.	7.6	183