

# Camille Charbonnier

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/11968264/publications.pdf>

Version: 2024-02-01

17  
papers

1,585  
citations

623734

14  
h-index

888059

17  
g-index

18  
all docs

18  
docs citations

18  
times ranked

3534  
citing authors

#	ARTICLE	IF	CITATIONS
1	Revisiting Li-Fraumeni Syndrome From <i>TP53</i> Mutation Carriers. <i>Journal of Clinical Oncology</i> , 2015, 33, 2345-2352.	1.6	525
2	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.	7.9	191
3	Phenotypic spectrum of probable and genetically-confirmed idiopathic basal ganglia calcification. <i>Brain</i> , 2013, 136, 3395-3407.	7.6	183
4	Contribution to Alzheimer's disease risk of rare variants in <i>TREM2</i> , <i>SORL1</i> , and <i>ABCA7</i> in 1779 cases and 1273 controls. <i>Neurobiology of Aging</i> , 2017, 59, 220.e1-220.e9.	3.1	116
5	Seizures in dominantly inherited Alzheimer disease. <i>Neurology</i> , 2016, 87, 912-919.	1.1	81
6	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, 710-716.	2.8	77
7	<i>SORL1</i> genetic variants and Alzheimer disease risk: a literature review and meta-analysis of sequencing data. <i>Acta Neuropathologica</i> , 2019, 138, 173-186.	7.7	77
8	Brain calcification process and phenotypes according to age and sex: Lessons from <i>SLC20A2</i> , <i>PDGFB</i> , and <i>PDGFRB</i> mutation carriers. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2015, 168, 586-594.	1.7	74
9	<i>ABCA7</i> rare variants and Alzheimer disease risk. <i>Neurology</i> , 2016, 86, 2134-2137.	1.1	63
10	Biallelic <i>MYORG</i> mutation carriers exhibit primary brain calcification with a distinct phenotype. <i>Brain</i> , 2019, 142, 1573-1586.	7.6	49
11	From Common to Rare Variants: The Genetic Component of Alzheimer Disease. <i>Human Heredity</i> , 2016, 81, 129-141.	0.8	37
12	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.	1.7	33
13	Contribution of genotoxic anticancer treatments to the development of multiple primary tumours in the context of germline <i>TP53</i> mutations. <i>European Journal of Cancer</i> , 2018, 101, 254-262.	2.8	32
14	Copy Number Variants in miR-138 as a Potential Risk Factor for Early-Onset Alzheimer's Disease. <i>Journal of Alzheimer's Disease</i> , 2019, 68, 1243-1255.	2.6	19
15	Germline Mutations of Inhibins in Early-Onset Ovarian Epithelial Tumors. <i>Human Mutation</i> , 2014, 35, 294-297.	2.5	11
16	Haploinsufficiency of the Primary Familial Brain Calcification Gene <i>SLC20A2</i> Mediated by Disruption of a Regulatory Element. <i>Movement Disorders</i> , 2020, 35, 1336-1345.	3.9	9
17	Penetrance estimation of Alzheimer disease in <i>SORL1</i> loss-of-function variant carriers using a family-based strategy and stratification by <i>APOE</i> genotypes. <i>Genome Medicine</i> , 2022, 14, .	8.2	7