Gianpiero L Cavalleri

List of Publications by Year
in descending order

139	13,104	41344	27406
papers	citations	49 h-index	106 g-index

156	156	156	19630
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	HLA-A*3101 and Carbamazepine-Induced Hypersensitivity Reactions in Europeans. New England Journal of Medicine, 2011, 364, 1134-1143.	27.0	815
2	Common genetic variants influence human subcortical brain structures. Nature, 2015, 520, 224-229.	27.8	772
3	Genomic insights into the origin of farming in the ancient Near East. Nature, 2016, 536, 419-424.	27.8	733
4	Natural selection on EPAS1 (HIF2Î±) associated with low hemoglobin concentration in Tibetan highlanders. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 11459-11464.	7.1	708
5	The ENIGMA Consortium: large-scale collaborative analyses of neuroimaging and genetic data. Brain Imaging and Behavior, 2014, 8, 153-182.	2.1	696
6	Identification of common variants associated with human hippocampal and intracranial volumes. Nature Genetics, 2012, 44, 552-561.	21.4	594
7	The genetic architecture of the human cerebral cortex. Science, 2020, 367, .	12.6	450
8	Structural brain abnormalities in the common epilepsies assessed in a worldwide ENIGMA study. Brain, 2018, 141, 391-408.	7.6	352
9	Genome-wide mega-analysis identifies 16 loci and highlights diverse biological mechanisms in the common epilepsies. Nature Communications, 2018, 9, 5269.	12.8	331
10	Genetic predictors of the maximum doses patients receive during clinical use of the anti-epileptic drugs carbamazepine and phenytoin. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 5507-5512.	7.1	321
11	Excavating Y-chromosome haplotype strata in Anatolia. Human Genetics, 2004, 114, 127-148.	3.8	318
12	Mapping cortical brain asymmetry in 17,141 healthy individuals worldwide via the ENIGMA Consortium. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E5154-E5163.	7.1	299
13	Genetic determinants of common epilepsies: a meta-analysis of genome-wide association studies. Lancet Neurology, The, 2014, 13, 893-903.	10.2	264
14	Novel genetic loci associated with hippocampal volume. Nature Communications, 2017, 8, 13624.	12.8	250
15	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. American Journal of Human Genetics, 2019, 105, 267-282.	6.2	237
16	Rare Deletions at 16p13.11 Predispose to a Diverse Spectrum of Sporadic Epilepsy Syndromes. American Journal of Human Genetics, 2010, 86, 707-718.	6.2	231
17	Novel genetic loci underlying human intracranial volume identified through genome-wide association. Nature Neuroscience, 2016, 19, 1569-1582.	14.8	213
18	Endothelial Nitric Oxide Synthase Gene Polymorphisms and Cardiovascular Disease: A HuGE Review. American Journal of Epidemiology, 2006, 164, 921-935.	3.4	210

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19	Genetic influences on schizophrenia and subcortical brain volumes: large-scale proof of concept. Nature Neuroscience, 2016, 19, 420-431.	14.8	204
20	Genetic architecture of subcortical brain structures in 38,851 individuals. Nature Genetics, 2019, 51, 1624-1636.	21.4	192
21	Ultra-rare genetic variation in common epilepsies: a case-control sequencing study. Lancet Neurology, The, 2017, 16, 135-143.	10.2	190
22	Multicentre search for genetic susceptibility loci in sporadic epilepsy syndrome and seizure types: a case-control study. Lancet Neurology, The, 2007, 6, 970-980.	10.2	175
23	Genetic Signatures Reveal High-Altitude Adaptation in a Set of Ethiopian Populations. Molecular Biology and Evolution, 2013, 30, 1877-1888.	8.9	173
24	Epilepsy, hippocampal sclerosis and febrile seizures linked by common genetic variation around SCN1A. Brain, 2013, 136, 3140-3150.	7.6	168
25	Personalized Medicine and Human Genetic Diversity. Cold Spring Harbor Perspectives in Medicine, 2014, 4, a008581-a008581.	6.2	140
26	The Y-Chromosome Tree Bursts into Leaf: 13,000 High-Confidence SNPs Covering the Majority of Known Clades. Molecular Biology and Evolution, 2015, 32, 661-673.	8.9	137
27	Common genetic variation and susceptibility to partial epilepsies: a genome-wide association study. Brain, 2010, 133, 2136-2147.	7.6	132
28	White matter abnormalities across different epilepsy syndromes in adults: an ENIGMA-Epilepsy study. Brain, 2020, 143, 2454-2473.	7.6	123
29	TDP2 protects transcription from abortive topoisomerase activity and is required for normal neural function. Nature Genetics, 2014, 46, 516-521.	21.4	122
30	Identifying Candidate Causal Variants Responsible for Altered Activity of the ABCB1 Multidrug Resistance Gene. Genome Research, 2004, 14, 1333-1344.	5.5	107
31	Understanding human diversity. Nature, 2005, 437, 1241-1242.	27.8	103
32	Distinct HLA associations of LGI1 and CASPR2-antibody diseases. Brain, 2018, 141, 2263-2271.	7.6	100
33	Tibetans living at sea level have a hyporesponsive hypoxia-inducible factor system and blunted physiological responses to hypoxia. Journal of Applied Physiology, 2014, 116, 893-904.	2.5	97
34	Network-based atrophy modeling in the common epilepsies: A worldwide ENIGMA study. Science Advances, 2020, 6, .	10.3	97
35	Exome Sequencing Followed by Large-Scale Genotyping Fails to Identify Single Rare Variants of Large Effect in Idiopathic Generalized Epilepsy. American Journal of Human Genetics, 2012, 91, 293-302.	6.2	95
36	A common polymorphism in the SCN1A gene associates with phenytoin serum levels at maintenance dose. Pharmacogenetics and Genomics, 2006, 16, 721-726.	1.5	93

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37	Polygenic burden in focal and generalized epilepsies. Brain, 2019, 142, 3473-3481.	7.6	90
38	Evolutionary history of Tibetans inferred from whole-genome sequencing. PLoS Genetics, 2017, 13, e1006675.	3.5	89
39	Failure to replicate previously reported genetic associations with sporadic temporal lobe epilepsy: where to from here?. Brain, 2005, 128, 1832-1840.	7.6	87
40	Role of SOX2 Mutations in Human Hippocampal Malformations and Epilepsy. Epilepsia, 2006, 47, 534-542.	5.1	85
41	A Multicenter Study of BRD2 as a Risk Factor for Juvenile Myoclonic Epilepsy. Epilepsia, 2007, 48, 706-712.	5.1	76
42	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. EBioMedicine, 2015, 2, 1063-1070.	6.1	74
43	Large-scale recent expansion of European patrilineages shown by population resequencing. Nature Communications, 2015, 6, 7152.	12.8	69
44	Rare coding variants in genes encoding GABAA receptors in genetic generalised epilepsies: an exome-based case-control study. Lancet Neurology, The, 2018, 17, 699-708.	10.2	67
45	The controversial association of ABCB1 polymorphisms in refractory epilepsy: An analysis of multiple SNPs in an Irish population. Epilepsy Research, 2007, 73, 192-198.	1.6	63
46	Ancient genomes from Iceland reveal the making of a human population. Science, 2018, 360, 1028-1032.	12.6	62
47	Different genetic components in the Norwegian population revealed by the analysis of mtDNA and Y chromosome polymorphisms. European Journal of Human Genetics, 2002, 10, 521-529.	2.8	55
48	Bayesian Meta-Analysis of Genetic Association Studies with Different Sets of Markers. American Journal of Human Genetics, 2008, 82, 859-872.	6.2	54
49	Carbamazepineâ€•and oxcarbazepineâ€induced hyponatremia in people with epilepsy. Epilepsia, 2017, 58, 1227-1233.	5.1	54
50	Association of Copy Number Variation of the 15q11.2 BP1-BP2 Region With Cortical and Subcortical Morphology and Cognition. JAMA Psychiatry, 2020, 77, 420.	11.0	54
51	Tackling Epilepsy With High-definition Precision Medicine. JAMA Neurology, 2019, 76, 1109.	9.0	53
52	Epilepsy in the mTORopathies: opportunities for precision medicine. Brain Communications, 2021, 3, fcab222.	3.3	53
53	The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield. Epilepsia, 2019, 60, 797-806.	5.1	52
54	Dose response of the 16p11.2 distal copy number variant on intracranial volume and basal ganglia. Molecular Psychiatry, 2020, 25, 584-602.	7.9	49

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55	Long- and short-term outcomes in renal allografts with deceased donors: A large recipient and donor genome-wide association study. American Journal of Transplantation, 2018, 18, 1370-1379.	4.7	47
56	The <scp>ENIGMAâ€Epilepsy</scp> working group: Mapping disease from large data sets. Human Brain Mapping, 2022, 43, 113-128.	3.6	47
57	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	7.6	47
58	Population structure and genome-wide patterns of variation in Ireland and Britain. European Journal of Human Genetics, 2010, 18, 1248-1254.	2.8	46
59	Vigabatrin Retinopathy in an Irish Cohort: Lack of Correlation with Dose. Epilepsia, 2006, 47, 311-317.	5.1	44
60	Genetic variation in CFH predicts phenytoin-induced maculopapular exanthema in European-descent patients. Neurology, 2018, 90, e332-e341.	1.1	43
61	SJS/TEN 2019: From science to translation. Journal of Dermatological Science, 2020, 98, 2-12.	1.9	41
62	Evaluating the causal relevance of diverse risk markers: horizontal systematic review. BMJ: British Medical Journal, 2009, 339, b4265-b4265.	2.3	40
63	A genomeâ€wide association study of recipient genotype and mediumâ€term kidney allograft function. Clinical Transplantation, 2013, 27, 379-387.	1.6	39
64	Genome-wide mapping for clinically relevant predictors of lamotrigine- and phenytoin-induced hypersensitivity reactions. Pharmacogenomics, 2012, 13, 399-405.	1.3	38
65	Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. American Journal of Human Genetics, 2021, 108, 965-982.	6.2	35
66	White matter alterations in patients with <scp>MRI</scp>â€negative temporal lobe epilepsy and their asymptomatic siblings. Epilepsia, 2015, 56, 1551-1561.	5.1	34
67	Comparative effectiveness of antiepileptic drugs in juvenile myoclonic epilepsy. Epilepsia Open, 2019, 4, 420-430.	2.4	34
68	Molecular variation of human HSP90Î± and HSP90Î² genes in Caucasians. Human Mutation, 2003, 21, 554-555.	2.5	33
69	No major role of common SV2A variation for predisposition or levetiracetam response in epilepsy. Epilepsy Research, 2009, 83, 44-51.	1.6	32
70	Temporal Cortex Morphology in Mesial Temporal Lobe Epilepsy Patients and Their Asymptomatic Siblings. Cerebral Cortex, 2016, 26, 1234-1241.	2.9	32
71	Pharmacogenomics and epilepsy: the road ahead. Pharmacogenomics, 2011, 12, 1429-1447.	1.3	31
72	Asymmetric cortical surface area and morphology changes in mesial temporal lobe epilepsy with hippocampal sclerosis. Epilepsia, 2012, 53, 995-1003.	5.1	31

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73	Genomic insights into the population structure and history of the Irish Travellers. Scientific Reports, 2017, 7, 42187.	3.3	31
74	Genomeâ€Wide Association Study Identifies Risk Loci for Cluster Headache. Annals of Neurology, 2021, 90, 193-202.	5.3	31
75	Genetic and Clinical Predictors of Age of ESKD in Individuals With Autosomal Dominant Tubulointerstitial Kidney Disease Due to UMOD Mutations. Kidney International Reports, 2020, 5, 1472-1485.	0.8	30
76	A comparison of genomic diagnostics in adults and children with epilepsy and comorbid intellectual disability. European Journal of Human Genetics, 2020, 28, 1066-1077.	2.8	30
77	Climate change and epilepsy: Insights from clinical and basic science studies. Epilepsy and Behavior, 2021, 116, 107791.	1.7	30
78	A cross-sectional MRI study of brain regional atrophy and clinical characteristics of temporal lobe epilepsy with hippocampal sclerosis. Epilepsy Research, 2012, 99, 156-166.	1.6	29
79	The Irish DNA Atlas: Revealing Fine-Scale Population Structure and History within Ireland. Scientific Reports, 2017, 7, 17199.	3.3	29
80	Genetic Variation in SENP1 and ANP32D as Predictors of Chronic Mountain Sickness. High Altitude Medicine and Biology, 2014, 15, 497-499.	0.9	28
81	Re-annotation of 191 developmental and epileptic encephalopathy-associated genes unmasks de novo variants in SCN1A. Npj Genomic Medicine, 2019, 4, 31.	3.8	27
82	A pharmacogenetic exploration of vigabatrin-induced visual field constriction. Epilepsy Research, 2006, 70, 144-152.	1.6	26
83	Weight change, genetics and antiepileptic drugs. Expert Review of Clinical Pharmacology, 2014, 7, 43-51.	3.1	26
84	Comparative effectiveness of antiepileptic drugs in patients with mesial temporal lobe epilepsy with hippocampal sclerosis. Epilepsia, 2017, 58, 1734-1741.	5.1	26
85	Polygenic risk score as a determinant of risk of non-melanoma skin cancer in a European-descent renal transplant cohort. American Journal of Transplantation, 2019, 19, 801-810.	4.7	26
86	The genetic landscape of Scotland and the Isles. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 19064-19070.	7.1	24
87	Testing association of rare genetic variants with resistance to three common antiseizure medications. Epilepsia, 2020, 61, 657-666.	5.1	22
88	A systemsâ€level analysis highlights microglial activation as a modifying factor in common epilepsies. Neuropathology and Applied Neurobiology, 2022, 48, .	3.2	22
89	Genetic structure in the Sherpa and neighboring Nepalese populations. BMC Genomics, 2017, 18, 102.	2.8	21
90	Autosomal dominant tubulointerstitial kidney disease (ADTKD) in Ireland. Renal Failure, 2019, 41, 832-841.	2.1	21

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91	Pharmacoresponse in genetic generalized epilepsy: a genome-wide association study. Pharmacogenomics, 2020, 21, 325-335.	1.3	21
92	Topographic divergence of atypical cortical asymmetry and atrophy patterns in temporal lobe epilepsy. Brain, 2022, 145, 1285-1298.	7.6	18
93	Examining the role of common genetic variation in the Î³2 subunit of the GABAA receptor in epilepsy using tagging SNPs. Epilepsy Research, 2006, 70, 229-238.	1.6	17
94	An Exome Sequencing Study of 10 Families with IgA Nephropathy. Nephron, 2020, 144, 72-83.	1.8	17
95	Genomic microdeletions associated with epilepsy: Not a contraindication to resective surgery. Epilepsia, 2011, 52, 1388-1392.	5.1	16
96	Heritability of Subcortical Volumetric Traits in Mesial Temporal Lobe Epilepsy. PLoS ONE, 2013, 8, e61880.	2.5	16
97	Population History and Altitude-Related Adaptation in the Sherpa. Frontiers in Physiology, 2019, 10, 1116.	2.8	16
98	Assessing the role of rare genetic variants in drugâ€resistant, nonâ€lesional focal epilepsy. Annals of Clinical and Translational Neurology, 2021, 8, 1376-1387.	3.7	16
99	Utility of Genomic Testing after Renal Biopsy. American Journal of Nephrology, 2020, 51, 43-53.	3.1	15
100	Exacerbation, then Clearance, of Mutation-Proven Darier's Disease of the Skin after Radiotherapy for Bronchial Carcinoma: A Case of Radiation-Induced Epidermal Differentiation?. Radiation Research, 2001, 156, 724-730.	1.5	14
101	MRIâ€Based Brain Structure Volumes in Temporal Lobe Epilepsy Patients and their Unaffected Siblings: A Preliminary Study. Journal of Neuroimaging, 2013, 23, 64-70.	2.0	14
102	The utility of a genetic kidney disease clinic employing a broad range of genomic testing platforms: experience of the Irish Kidney Gene Project. Journal of Nephrology, 2022, 35, 1655-1665.	2.0	14
103	Revealing the recent demographic history of Europe via haplotype sharing in the UK Biobank. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	7.1	14
104	The impact of donor and recipient common clinical and genetic variation on estimated glomerular filtration rate in a European renal transplant population. American Journal of Transplantation, 2019, 19, 2262-2273.	4.7	13
105	Genomic and clinical predictors of lacosamide response in refractory epilepsies. Epilepsia Open, 2019, 4, 563-571.	2.4	12
106	Exploring the genetic overlap between psychiatric illness and epilepsy: A review. Epilepsy and Behavior, 2020, 102, 106669.	1.7	12
107	The impact of ERBB-family germline single nucleotide polymorphisms on survival response to adjuvant trastuzumab treatment in HER2-positive breast cancer. Oncotarget, 2016, 7, 75518-75525.	1.8	12
108	The role of common genetic variation in presumed monogenic epilepsies. EBioMedicine, 2022, 81, 104098.	6.1	12

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109	Polygenic risk score of nonâ€melanoma skin cancer predicts postâ€transplant skin cancer across multiple organ types. Clinical Transplantation, 2020, 34, e13904.	1.6	11
110	The genetic landscape of polycystic kidney disease in Ireland. European Journal of Human Genetics, 2021, 29, 827-838.	2.8	11
111	Postictal Psychosis in Epilepsy: A Clinicogenetic Study. Annals of Neurology, 2021, 90, 464-476.	5.3	11
112	A pharmacogenomic assessment of psychiatric adverse drug reactions to levetiracetam. Epilepsia, 2022, 63, 1563-1570.	5.1	11
113	Opportunities and Challenges for Genome Sequencing in the Clinic. Advances in Protein Chemistry and Structural Biology, 2012, 89, 65-83.	2.3	9
114	Association of CYP3A variants with kidney transplant outcomes. Renal Failure, 2015, 37, 562-566.	2.1	9
115	Genomics-Guided Precise Anti-Epileptic Drug Development. Neurochemical Research, 2017, 42, 2084-2088.	3.3	9
116	A genomeâ€wide association study of sodium levels and drug metabolism in an epilepsy cohort treated with carbamazepine and oxcarbazepine. Epilepsia Open, 2019, 4, 102-109.	2.4	9
117	De-novo mutations in patients with chronic ultra-refractory epilepsy with onset after age five years. European Journal of Medical Genetics, 2020, 63, 103625.	1.3	9
118	Using common genetic variants to find drugs for common epilepsies. Brain Communications, 2021, 3, fcab287.	3.3	9
119	Educating pharmacy students through a pandemic: Reflecting on our COVID-19 experience. Research in Social and Administrative Pharmacy, 2021, , .	3.0	8
120	Concordance between PCR-based extraction-free saliva and nasopharyngeal swabs for SARS-CoV-2 testing. HRB Open Research, 0, 4, 85.	0.6	8
121	Association of ultraâ€rare coding variants with genetic generalized epilepsy: A caseâ€“control whole exome sequencing study. Epilepsia, 2022, 63, 723-735.	5.1	8
122	Genome-Scale Methods Converge on Key Mitochondrial Genes for the Survival of Human Cardiomyocytes in Hypoxia. Circulation: Cardiovascular Genetics, 2014, 7, 407-415.	5.1	7
123	Two Further Blood Pressure Loci Identified in Ion Channel Genes With a Genecentric Approach. Circulation: Cardiovascular Genetics, 2014, 7, 873-879.	5.1	7
124	Development of a genomics module within an epilepsyâ€specific electronic health record: Toward genomic medicine in epilepsy care. Epilepsia, 2019, 60, 1670-1677.	5.1	7
125	Normal cerebral cortical thickness in first-degree relatives of temporal lobe epilepsy patients. Neurology, 2019, 92, e351-e358.	1.1	7
126	Concordance between PCR-based extraction-free saliva and nasopharyngeal swabs for SARS-CoV-2 testing. HRB Open Research, 2021, 4, 85.	0.6	7

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127	Diagnostic utility of genetic testing in patients undergoing renal biopsy. Journal of Physical Education and Sports Management, 2020, 6, a005462.	1.2	7
128	Role of Common Genetic Variants for Drug-Resistance to Specific Anti-Seizure Medications. Frontiers in Pharmacology, 2021, 12, 688386.	3.5	6
129	An assessment of the Irish population for large-scale genetic mapping studies involving epilepsy and other complex diseases. European Journal of Human Genetics, 2008, 16, 176-183.	2.8	5
130	Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations. Epilepsia, 2021, 62, 1518-1527.	5.1	5
131	The genetic determinants of renal allograft rejection. American Journal of Transplantation, 2018, 18, 2100-2101.	4.7	4
132	A Rare Autosomal Dominant Variant in Regulator of Calcineurin Type 1 (RCAN1) Gene Confers Enhanced Calcineurin Activity and May Cause FSGS. Journal of the American Society of Nephrology: JASN, 2021, 32, 1682-1695.	6.1	3
133	Genomic analysis of â€œmicrophenotypesâ€•in epilepsy. American Journal of Medical Genetics, Part A, 2022, 188, 138-146.	1.2	3
134	eHealth as a Facilitator of Precision Medicine in Epilepsy. Biomedicine Hub, 2017, 2, 1-9.	1.2	2
135	Renal transplant outcomes in patients with autosomal dominant tubulointerstitial kidney disease. Clinical Transplantation, 2020, 34, e13783.	1.6	2
136	Response 2 to pharmacogenetic screening to prevent carbamazepine-induced toxic epidermal necrolysis and Stevens-Johnson syndrome: a critical appraisal. British Journal of Dermatology, 2012, 166, 12-14.	1.5	1
137	The relationship between donor-recipient genetic distance and long-term kidney transplant outcome. HRB Open Research, 2020, 3, 47.	0.6	1
138	Population diversity, genomes and disease. , 0, , 80-91.		0
139	Whole exome sequencing studies in epilepsy: A deep analysis of the published literature. American Journal of Medical Genetics, Part A, 2022, , .	1.2	0