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	The <scp>ENIGMAâ€Epilepsy</scp> working group: Mapping disease from large data sets. Human Brain Mapping, 2022, 43, 113-128.	3.6	47
2	A systemsâ€level analysis highlights microglial activation as a modifying factor in common epilepsies. Neuropathology and Applied Neurobiology, 2022, 48, .	3.2	22
3	Genomic analysis of â€œmicrophenotypesâ€in epilepsy. American Journal of Medical Genetics, Part A, 2022, 188, 138-146.	1.2	3
4	Topographic divergence of atypical cortical asymmetry and atrophy patterns in temporal lobe epilepsy. Brain, 2022, 145, 1285-1298.	7.6	18
5	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
6	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
7	Whole exome sequencing studies in epilepsy: A deep analysis of the published literature. American Journal of Medical Genetics, Part A, 2022, , .	1.2	0
8	A pharmacogenomic assessment of psychiatric adverse drug reactions to levetiracetam. Epilepsia, 2022, 63, 1563-1570.	5.1	11
9	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
10	The role of common genetic variation in presumed monogenic epilepsies. EBioMedicine, 2022, 81, 104098.	6.1	12
11	The genetic landscape of polycystic kidney disease in Ireland. European Journal of Human Genetics, 2021, 29, 827-838.	2.8	11
12	Climate change and epilepsy: Insights from clinical and basic science studies. Epilepsy and Behavior, 2021, 116, 107791.	1.7	30
13	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
14	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
15	Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations. Epilepsia, 2021, 62, 1518-1527.	5.1	5
16	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
17	Role of Common Genetic Variants for Drug-Resistance to Specific Anti-Seizure Medications. Frontiers in Pharmacology, 2021, 12, 688386.	3.5	6
18	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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19	Genomeâ€Wide Association Study Identifies Risk Loci for Cluster Headache. Annals of Neurology, 2021, 90, 193-202.	5.3	31
20	Educating pharmacy students through a pandemic: Reflecting on our COVID-19 experience. Research in Social and Administrative Pharmacy, 2021, , .	3.0	8
21	Postictal Psychosis in Epilepsy: A Clinicogenetic Study. Annals of Neurology, 2021, 90, 464-476.	5.3	11
22	Epilepsy in the mTORopathies: opportunities for precision medicine. Brain Communications, 2021, 3, fcab222.	3.3	53
23	Using common genetic variants to find drugs for common epilepsies. Brain Communications, 2021, 3, fcab287.	3.3	9
24	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
25	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
26	Utility of Genomic Testing after Renal Biopsy. American Journal of Nephrology, 2020, 51, 43-53.	3.1	15
27	Exploring the genetic overlap between psychiatric illness and epilepsy: A review. Epilepsy and Behavior, 2020, 102, 106669.	1.7	12
28	An Exome Sequencing Study of 10 Families with IgA Nephropathy. Nephron, 2020, 144, 72-83.	1.8	17
29	Network-based atrophy modeling in the common epilepsies: A worldwide ENIGMA study. Science Advances, 2020, 6, .	10.3	97
30	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
31	White matter abnormalities across different epilepsy syndromes in adults: an ENIGMA-Epilepsy study. Brain, 2020, 143, 2454-2473.	7.6	123
32	Pharmacoresponse in genetic generalized epilepsy: a genome-wide association study. Pharmacogenomics, 2020, 21, 325-335.	1.3	21
33	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
34	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	7.6	47
35	Testing association of rare genetic variants with resistance to three common antiseizure medications. Epilepsia, 2020, 61, 657-666.	5.1	22
36	SJS/TEN 2019: From science to translation. Journal of Dermatological Science, 2020, 98, 2-12.	1.9	41

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37	The genetic architecture of the human cerebral cortex. Science, 2020, 367, .	12.6	450
38	Renal transplant outcomes in patients with autosomal dominant tubulointerstitial kidney disease. Clinical Transplantation, 2020, 34, e13783.	1.6	2
39	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
40	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
41	The relationship between donor-recipient genetic distance and long-term kidney transplant outcome. HRB Open Research, 2020, 3, 47.	0.6	1
42	Diagnostic utility of genetic testing in patients undergoing renal biopsy. Journal of Physical Education and Sports Management, 2020, 6, a005462.	1.2	7
43	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
44	Tackling Epilepsy With High-definition Precision Medicine. JAMA Neurology, 2019, 76, 1109.	9.0	53
45	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
46	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
47	Comparative effectiveness of antiepileptic drugs in juvenile myoclonic epilepsy. Epilepsia Open, 2019, 4, 420-430.	2.4	34
48	Polygenic burden in focal and generalized epilepsies. Brain, 2019, 142, 3473-3481.	7.6	90
49	Genomic and clinical predictors of lacosamide response in refractory epilepsies. Epilepsia Open, 2019, 4, 563-571.	2.4	12
50	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
51	Autosomal dominant tubulointerstitial kidney disease (ADTKD) in Ireland. Renal Failure, 2019, 41, 832-841.	2.1	21
52	Population History and Altitude-Related Adaptation in the Sherpa. Frontiers in Physiology, 2019, 10, 1116.	2.8	16
53	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
54	The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield. Epilepsia, 2019, 60, 797-806.	5.1	52

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55	Genetic architecture of subcortical brain structures in 38,851 individuals. Nature Genetics, 2019, 51, 1624-1636.	21.4	192
56	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
57	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
58	Normal cerebral cortical thickness in first-degree relatives of temporal lobe epilepsy patients. Neurology, 2019, 92, e351-e358.	1.1	7
59	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
60	Structural brain abnormalities in the common epilepsies assessed in a worldwide ENIGMA study. Brain, 2018, 141, 391-408.	7.6	352
61	Genetic variation in CFH predicts phenytoin-induced maculopapular exanthema in European-descent patients. Neurology, 2018, 90, e332-e341.	1.1	43
62	The genetic determinants of renal allograft rejection. American Journal of Transplantation, 2018, 18, 2100-2101.	4.7	4
63	Genome-wide mega-analysis identifies 16 loci and highlights diverse biological mechanisms in the common epilepsies. Nature Communications, 2018, 9, 5269.	12.8	331
64	Distinct HLA associations of LGI1 and CASPR2-antibody diseases. Brain, 2018, 141, 2263-2271.	7.6	100
65	Ancient genomes from Iceland reveal the making of a human population. Science, 2018, 360, 1028-1032.	12.6	62
66	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
67	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
68	Novel genetic loci associated with hippocampal volume. Nature Communications, 2017, 8, 13624.	12.8	250
69	Ultra-rare genetic variation in common epilepsies: a case-control sequencing study. Lancet Neurology, The, 2017, 16, 135-143.	10.2	190
70	Genomic insights into the population structure and history of the Irish Travellers. Scientific Reports, 2017, 7, 42187.	3.3	31
71	Genomics-Guided Precise Anti-Epileptic Drug Development. Neurochemical Research, 2017, 42, 2084-2088.	3.3	9
72	Carbamazepineâ€and oxcarbazepineâ€induced hyponatremia in people with epilepsy. Epilepsia, 2017, 58, 1227-1233.	5.1	54

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73	Comparative effectiveness of antiepileptic drugs in patients with mesial temporal lobe epilepsy with hippocampal sclerosis. Epilepsia, 2017, 58, 1734-1741.	5.1	26
74	The Irish DNA Atlas: Revealing Fine-Scale Population Structure and History within Ireland. Scientific Reports, 2017, 7, 17199.	3.3	29
75	Genetic structure in the Sherpa and neighboring Nepalese populations. BMC Genomics, 2017, 18, 102.	2.8	21
76	eHealth as a Facilitator of Precision Medicine in Epilepsy. Biomedicine Hub, 2017, 2, 1-9.	1.2	2
77	Evolutionary history of Tibetans inferred from whole-genome sequencing. PLoS Genetics, 2017, 13, e1006675.	3.5	89
78	Novel genetic loci underlying human intracranial volume identified through genome-wide association. Nature Neuroscience, 2016, 19, 1569-1582.	14.8	213
79	Genomic insights into the origin of farming in the ancient Near East. Nature, 2016, 536, 419-424.	27.8	733
80	Genetic influences on schizophrenia and subcortical brain volumes: large-scale proof of concept. Nature Neuroscience, 2016, 19, 420-431.	14.8	204
81	Temporal Cortex Morphology in Mesial Temporal Lobe Epilepsy Patients and Their Asymptomatic Siblings. Cerebral Cortex, 2016, 26, 1234-1241.	2.9	32
82	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
83	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. EBioMedicine, 2015, 2, 1063-1070.	6.1	74
84	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
85	Large-scale recent expansion of European patrilineages shown by population resequencing. Nature Communications, 2015, 6, 7152.	12.8	69
86	Association of CYP3A variants with kidney transplant outcomes. Renal Failure, 2015, 37, 562-566.	2.1	9
87	Common genetic variants influence human subcortical brain structures. Nature, 2015, 520, 224-229.	27.8	772
88	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
89	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
90	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

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91	Two Further Blood Pressure Loci Identified in Ion Channel Genes With a Genecentric Approach. Circulation: Cardiovascular Genetics, 2014, 7, 873-879.	5.1	7
92	TDP2 protects transcription from abortive topoisomerase activity and is required for normal neural function. Nature Genetics, 2014, 46, 516-521.	21.4	122
93	The ENIGMA Consortium: large-scale collaborative analyses of neuroimaging and genetic data. Brain Imaging and Behavior, 2014, 8, 153-182.	2.1	696
94	Genetic Variation in SENP1 and ANP32D as Predictors of Chronic Mountain Sickness. High Altitude Medicine and Biology, 2014, 15, 497-499.	0.9	28
95	Personalized Medicine and Human Genetic Diversity. Cold Spring Harbor Perspectives in Medicine, 2014, 4, a008581-a008581.	6.2	140
96	Genetic determinants of common epilepsies: a meta-analysis of genome-wide association studies. Lancet Neurology, The, 2014, 13, 893-903.	10.2	264
97	Weight change, genetics and antiepileptic drugs. Expert Review of Clinical Pharmacology, 2014, 7, 43-51.	3.1	26
98	A genomeâ€wide association study of recipient genotype and mediumâ€term kidney allograft function. Clinical Transplantation, 2013, 27, 379-387.	1.6	39
99	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
100	Epilepsy, hippocampal sclerosis and febrile seizures linked by common genetic variation around SCN1A. Brain, 2013, 136, 3140-3150.	7.6	168
101	Genetic Signatures Reveal High-Altitude Adaptation in a Set of Ethiopian Populations. Molecular Biology and Evolution, 2013, 30, 1877-1888.	8.9	173
102	Heritability of Subcortical Volumetric Traits in Mesial Temporal Lobe Epilepsy. PLoS ONE, 2013, 8, e61880.	2.5	16
103	Genome-wide mapping for clinically relevant predictors of lamotrigine- and phenytoin-induced hypersensitivity reactions. Pharmacogenomics, 2012, 13, 399-405.	1.3	38
104	Opportunities and Challenges for Genome Sequencing in the Clinic. Advances in Protein Chemistry and Structural Biology, 2012, 89, 65-83.	2.3	9
105	Identification of common variants associated with human hippocampal and intracranial volumes. Nature Genetics, 2012, 44, 552-561.	21.4	594
106	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
107	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
108	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

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109	Asymmetric cortical surface area and morphology changes in mesial temporal lobe epilepsy with hippocampal sclerosis. Epilepsia, 2012, 53, 995-1003.	5.1	31
110	HLA-A*3101 and Carbamazepine-Induced Hypersensitivity Reactions in Europeans. New England Journal of Medicine, 2011, 364, 1134-1143.	27.0	815
111	Genomic microdeletions associated with epilepsy: Not a contraindication to resective surgery. Epilepsia, 2011, 52, 1388-1392.	5.1	16
112	Pharmacogenomics and epilepsy: the road ahead. Pharmacogenomics, 2011, 12, 1429-1447.	1.3	31
113	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
114	Population structure and genome-wide patterns of variation in Ireland and Britain. European Journal of Human Genetics, 2010, 18, 1248-1254.	2.8	46
115	Common genetic variation and susceptibility to partial epilepsies: a genome-wide association study. Brain, 2010, 133, 2136-2147.	7.6	132
116	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
117	Evaluating the causal relevance of diverse risk markers: horizontal systematic review. BMJ: British Medical Journal, 2009, 339, b4265-b4265.	2.3	40
118	No major role of common SV2A variation for predisposition or levetiracetam response in epilepsy. Epilepsy Research, 2009, 83, 44-51.	1.6	32
119	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
120	Bayesian Meta-Analysis of Genetic Association Studies with Different Sets of Markers. American Journal of Human Genetics, 2008, 82, 859-872.	6.2	54
121	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
122	A Multicenter Study of BRD2 as a Risk Factor for Juvenile Myoclonic Epilepsy. Epilepsia, 2007, 48, 706-712.	5.1	76
123	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
124	Endothelial Nitric Oxide Synthase Gene Polymorphisms and Cardiovascular Disease: A HuGE Review. American Journal of Epidemiology, 2006, 164, 921-935.	3.4	210
125	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
126	Vigabatrin Retinopathy in an Irish Cohort: Lack of Correlation with Dose. Epilepsia, 2006, 47, 311-317.	5.1	44

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127	Role of SOX2 Mutations in Human Hippocampal Malformations and Epilepsy. Epilepsia, 2006, 47, 534-542.	5.1	85
128	A pharmacogenetic exploration of vigabatrin-induced visual field constriction. Epilepsy Research, 2006, 70, 144-152.	1.6	26
129	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
130	Understanding human diversity. Nature, 2005, 437, 1241-1242.	27.8	103
131	Failure to replicate previously reported genetic associations with sporadic temporal lobe epilepsy: where to from here?. Brain, 2005, 128, 1832-1840.	7.6	87
132	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
133	Identifying Candidate Causal Variants Responsible for Altered Activity of the ABCB1 Multidrug Resistance Gene. Genome Research, 2004, 14, 1333-1344.	5.5	107
134	Excavating Y-chromosome haplotype strata in Anatolia. Human Genetics, 2004, 114, 127-148.	3.8	318
135	Molecular variation of human HSP90Î± and HSP90Î² genes in Caucasians. Human Mutation, 2003, 21, 554-555.	2.5	33
136	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
137	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
138	Population diversity, genomes and disease. , 0, , 80-91.		0
139	Concordance between PCR-based extraction-free saliva and nasopharyngeal swabs for SARS-CoV-2 testing. HRB Open Research, 0, 4, 85.	0.6	8