SlavÃ© Petrovski

List of Publications by Year
in descending order

99 papers	11,530 citations	47 h-index	99 g-index
		47006	33894
117 all docs	117 docs citations	117 times ranked	19352 citing authors

#	Article	IF	Citations
1	De novo mutations in epileptic encephalopathies. Nature, 2013, 501, 217-221.	27.8	1,351
2	Genic Intolerance to Functional Variation and the Interpretation of Personal Genomes. PLoS Genetics, 2013, 9, e1003709.	3.5	844
3	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. Science, 2015, 347, 1436-1441.	12.6	823
4	Diagnostic Utility of Exome Sequencing for Kidney Disease. New England Journal of Medicine, 2019, 380, 142-151.	27.0	456
5	De Novo Mutations in Synaptic Transmission Genes Including DNM1 Cause Epileptic Encephalopathies. American Journal of Human Genetics, 2014, 95, 360-370.	6.2	388
6	Whole-exome sequencing in the evaluation of fetal structural anomalies: a prospective cohort study. Lancet, The, 2019, 393, 758-767.	13.7	368
7	Whole-exome sequencing in undiagnosed genetic diseases: interpreting 119 trios. Genetics in Medicine, 2015, 17, 774-781.	2.4	284
8	Genetic determinants of common epilepsies: a meta-analysis of genome-wide association studies. Lancet Neurology, The, 2014, 13, 893-903.	10.2	264
9	KCNT1 gain of function in 2 epilepsy phenotypes is reversed by quinidine. Annals of Neurology, 2014, 75, 581-590.	5.3	249
10	De Novo Mutations in SLC1A2 and CACNA1A Are Important Causes of Epileptic Encephalopathies. American Journal of Human Genetics, 2016, 99, 287-298.	6.2	247
11	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
12	Sequencing studies in human genetics: design and interpretation. Nature Reviews Genetics, 2013, 14, 460-470.	16.3	236
13	Advancing human genetics research and drug discovery through exome sequencing of the UK Biobank. Nature Genetics, 2021, 53, 942-948.	21.4	234
14	Rare variant contribution to human disease in 281,104 UK Biobank exomes. Nature, 2021, 597, 527-532.	27.8	224
15	Exomeâ€based analysis of cardiac arrhythmia, respiratory control, and epilepsy genes in sudden unexpected death in epilepsy. Annals of Neurology, 2016, 79, 522-534.	5.3	216
16	Unequal representation of genetic variation across ancestry groups creates healthcare inequalityÂin the application of precision medicine. Genome Biology, 2016, 17, 157.	8.8	198
17	An Exome Sequencing Study to Assess the Role of Rare Genetic Variation in Pulmonary Fibrosis. American Journal of Respiratory and Critical Care Medicine, 2017, 196, 82-93.	5.6	185
18	Quinidine in the treatment of <scp>KCNT</scp>1â€positive epilepsies. Annals of Neurology, 2015, 78, 995-999.	5.3	184

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19	Inherited DOCK2 Deficiency in Patients with Early-Onset Invasive Infections. New England Journal of Medicine, 2015, 372, 2409-2422.	27.0	169
20	Epilepsy, hippocampal sclerosis and febrile seizures linked by common genetic variation around SCN1A. Brain, 2013, 136, 3140-3150.	7.6	168
21	Systems genetics identifies Sestrin 3 as a regulator of a proconvulsant gene network in human epileptic hippocampus. Nature Communications, 2015, 6, 6031.	12.8	158
22	Mechanistic Insight into NMDA Receptor Dysregulation by Rare Variants in the GluN2A and GluN2B Agonist Binding Domains. American Journal of Human Genetics, 2016, 99, 1261-1280.	6.2	158
23	Optimizing genomic medicine in epilepsy through a gene-customized approach to missense variant interpretation. Genome Research, 2017, 27, 1715-1729.	5.5	150
24	Rare-variant collapsing analyses for complex traits: guidelines and applications. Nature Reviews Genetics, 2019, 20, 747-759.	16.3	147
25	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136
26	Systems genetics identifies a convergent gene network for cognition and neurodevelopmental disease. Nature Neuroscience, 2016, 19, 223-232.	14.8	131
27	One gene, many neuropsychiatric disorders: lessons from Mendelian diseases. Nature Neuroscience, 2014, 17, 773-781.	14.8	128
28	The Intolerance of Regulatory Sequence to Genetic Variation Predicts Gene Dosage Sensitivity. PLoS Genetics, 2015, 11, e1005492.	3.5	123
29	Annotating pathogenic non-coding variants in genic regions. Nature Communications, 2017, 8, 236.	12.8	122
30	The intolerance to functional genetic variation of protein domains predicts the localization of pathogenic mutations within genes. Genome Biology, 2016, 17, 9.	8.8	118
31	Molecular Mechanism of Disease-Associated Mutations in the Pre-M1 Helix of NMDA Receptors and Potential Rescue Pharmacology. PLoS Genetics, 2017, 13, e1006536.	3.5	117
32	Wholeâ€exome sequencing in 20,197 persons for rare variants in Alzheimer's disease. Annals of Clinical and Translational Neurology, 2018, 5, 832-842.	3.7	112
33	Genome-wide analyses of 200,453 individuals yield new insights into the causes and consequences of clonal hematopoiesis. Nature Genetics, 2022, 54, 1155-1166.	21.4	109
34	Germline De Novo Mutations in GNB1 Cause Severe Neurodevelopmental Disability, Hypotonia, and Seizures. American Journal of Human Genetics, 2016, 98, 1001-1010.	6.2	102
35	A Genome-wide Comparison of the Functional Properties of Rare and Common Genetic Variants in Humans. American Journal of Human Genetics, 2011, 88, 458-468.	6.2	89
36	De Novo Mutations in SON Disrupt RNA Splicing of Genes Essential for Brain Development and Metabolism, Causing an Intellectual-Disability Syndrome. American Journal of Human Genetics, 2016, 99, 711-719.	6.2	81

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37	Genomewide Association Study for Determinants of HIV-1 Acquisition and Viral Set Point in HIV-1 Serodiscordant Couples with Quantified Virus Exposure. PLoS ONE, 2011, 6, e28632.	2.5	80
38	Common human genetic variants and HIV-1 susceptibility: a genome-wide survey in a homogeneous African population. Aids, 2011, 25, 513-518.	2.2	77
39	Epilepsy genetics: clinical impacts and biological insights. Lancet Neurology, The, 2020, 19, 93-100.	10.2	75
40	Pan-ancestry exome-wide association analyses of COVID-19 outcomes in 586,157 individuals. American Journal of Human Genetics, 2021, 108, 1350-1355.	6.2	72
41	Mosaic mutations in early-onset genetic diseases. Genetics in Medicine, 2016, 18, 746-749.	2.4	70
42	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 991-999.	6.2	68
43	Lung Transplant Outcomes in Patients With Pulmonary Fibrosis With Telomere-Related Gene Variants. Chest, 2019, 156, 477-485.	0.8	60
44	Copy number variant analysis from exome data in 349 patients with epileptic encephalopathy. Annals of Neurology, 2015, 78, 323-328.	5.3	59
45	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. American Journal of Human Genetics, 2019, 104, 164-178.	6.2	59
46	Dominant Splice Site Mutations in PIK3R1 Cause Hyper IgM Syndrome, Lymphadenopathy and Short Stature. Journal of Clinical Immunology, 2016, 36, 462-471.	3.8	55
47	The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield. Epilepsia, 2019, 60, 797-806.	5.1	52
48	De novoGRINvariants in NMDA receptor M2 channel poreâ€forming loop are associated with neurological diseases. Human Mutation, 2019, 40, 2393-2413.	2.5	48
49	Refining the phenotype associated with GNB1 mutations: Clinical data on 18 newly identified patients and review of the literature. American Journal of Medical Genetics, Part A, 2018, 176, 2259-2275.	1.2	47
50	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	7.6	47
51	Epileptic encephalopathy-causing mutations in DNM1 impair synaptic vesicle endocytosis. Neurology: Genetics, 2015, 1, e4.	1.9	46
52	De Novo Heterozygous POLR2A Variants Cause a Neurodevelopmental Syndrome with Profound Infantile-Onset Hypotonia. American Journal of Human Genetics, 2019, 105, 283-301.	6.2	46
53	MTR-Viewer: identifying regions within genes under purifying selection. Nucleic Acids Research, 2019, 47, W121-W126.	14.5	43
54	Spontaneous Coronary Artery Dissection. Circulation Genomic and Precision Medicine, 2020, 13, e003030.	3.6	43

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55	Timeâ€frequency mapping of the rhythmic limb movements distinguishes convulsive epileptic from psychogenic nonepileptic seizures. Epilepsia, 2013, 54, 1402-1408.	5.1	42
56	Exome-Based Rare-Variant Analyses in CKD. Journal of the American Society of Nephrology: JASN, 2019, 30, 1109-1122.	6.1	40
57	Prioritizing non-coding regions based on human genomic constraint and sequence context with deep learning. Nature Communications, 2021, 12, 1504.	12.8	40
58	Multidrugâ€resistant genotype (ABCB1) and seizure recurrence in newly treated epilepsy: Data from international pharmacogenetic cohorts. Epilepsia, 2009, 50, 1689-1696.	5.1	39
59	The importance of dynamic re-analysis in diagnostic whole exome sequencing. Journal of Medical Genetics, 2017, 54, 155-156.	3.2	38
60	ExACtly zero or once. Neurology: Genetics, 2017, 3, e163.	1.9	37
61	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
62	Inhibition of microRNA 128 promotes excitability of cultured cortical neuronal networks. Genome Research, 2016, 26, 1411-1416.	5.5	34
63	Comparative effectiveness of antiepileptic drugs in juvenile myoclonic epilepsy. Epilepsia Open, 2019, 4, 420-430.	2.4	34
64	A genome-wide association study and biological pathway analysis of epilepsy prognosis in a prospective cohort of newly treated epilepsy. Human Molecular Genetics, 2014, 23, 247-258.	2.9	33
65	De Novo Missense Variants in FBXW11 Cause Diverse Developmental Phenotypes Including Brain, Eye, and Digit Anomalies. American Journal of Human Genetics, 2019, 105, 640-657.	6.2	31
66	Orion: Detecting regions of the human non-coding genome that are intolerant to variation using population genetics. PLoS ONE, 2017, 12, e0181604.	2.5	31
67	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	6.2	30
68	Incorporating Functional Information in Tests of Excess De Novo Mutational Load. American Journal of Human Genetics, 2015, 97, 272-283.	6.2	29
69	A comprehensive approach to identifying repurposed drugs to treat <scp>SCN</scp>8A epilepsy. Epilepsia, 2018, 59, 802-813.	5.1	29
70	Gain-of-functionHCN2variants in genetic epilepsy. Human Mutation, 2018, 39, 202-209.	2.5	28
71	Identification of a missense variant in SPDL1 associated with idiopathic pulmonary fibrosis. Communications Biology, 2021, 4, 392.	4.4	28
72	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

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73	Rare and Common Variants in KIF15 Contribute to Genetic Risk of Idiopathic Pulmonary Fibrosis. American Journal of Respiratory and Critical Care Medicine, 2022, 206, 56-69.	5.6	25
74	Exome sequencing results in successful riboflavin treatment of a rapidly progressive neurological condition. Journal of Physical Education and Sports Management, 2015, 1, a000257.	1.2	24
75	Mantis-ml: Disease-Agnostic Gene Prioritization from High-Throughput Genomic Screens by Stochastic Semi-supervised Learning. American Journal of Human Genetics, 2020, 106, 659-678.	6.2	24
76	Multi-SNP pharmacogenomic classifier is superior to single-SNP models for predicting drug outcome in complex diseases. Pharmacogenetics and Genomics, 2009, 19, 147-152.	1.5	23
77	Substitution Monotherapy With Levetiracetam vs Older Antiepileptic Drugs. Archives of Neurology, 2012, 69, 1563.	4.5	23
78	meaRtools: An R package for the analysis of neuronal networks recorded on microelectrode arrays. PLoS Computational Biology, 2018, 14, e1006506.	3.2	22
79	A new approach for rare variation collapsing on functional protein domains implicates specific genic regions in ALS. Genome Research, 2019, 29, 809-818.	5.5	21
80	Lossâ€ofâ€function variants in NFIA provide further support that NFIA is a critical gene in 1p32â€p31 deletion syndrome: A four patient series. American Journal of Medical Genetics, Part A, 2017, 173, 3158-3164.	1.2	16
81	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
82	Evaluating whole genome sequence data from the Genetic Absence Epilepsy Rat from Strasbourg and its related non-epileptic strain. PLoS ONE, 2017, 12, e0179924.	2.5	16
83	Validation of a multigenic model to predict seizure control in newly treated epilepsy. Epilepsy Research, 2014, 108, 1797-1805.	1.6	15
84	Unraveling the genetics of common epilepsies: Approaches, platforms, and caveats. Epilepsy and Behavior, 2013, 26, 229-233.	1.7	14
85	Phenomics and the Interpretation of Personal Genomes. Science Translational Medicine, 2014, 6, 254fs35.	12.4	13
86	Association of SLC32A1 Missense Variants With Genetic Epilepsy With Febrile Seizures Plus. Neurology, 2021, 96, e2251-e2260.	1.1	13
87	Diverse genetic causes of polymicrogyria with epilepsy. Epilepsia, 2021, 62, 973-983.	5.1	12
88	Sustained therapeutic response to riboflavin in a child with a progressive neurological condition, diagnosed by whole-exome sequencing. Journal of Physical Education and Sports Management, 2015, 1, a000265.	1.2	11
89	Antiepileptic Drug Teratogenicity and De Novo Genetic Variation Load. Annals of Neurology, 2020, 87, 897-906.	5.3	9
90	Using common genetic variants to find drugs for common epilepsies. Brain Communications, 2021, 3, fcab287.	3.3	9

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91	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
92	Is FGF13 a major contributor to genetic epilepsy with febrile seizures plus?. Epilepsy Research, 2016, 128, 48-51.	1.6	7
93	Development of a rapid functional assay that predicts GLUT1 disease severity. Neurology: Genetics, 2018, 4, e297.	1.9	7
94	Potential role of regulatory DNA variants in modifying the risk of severe cutaneous reactions induced by aromatic antiâ€seizure medications. Epilepsia, 2022, 63, 936-949.	5.1	5
95	Gene-SCOUT: identifying genes with similar continuous trait fingerprints from phenome-wide association analyses. Nucleic Acids Research, 2022, 50, 4289-4301.	14.5	3
96	eHealth as a Facilitator of Precision Medicine in Epilepsy. Biomedicine Hub, 2017, 2, 1-9.	1.2	2
97	Uncovering variable neoplasms between <scp>ATM</scp> proteinâ€truncating and common missense variants using 394â€‰694 <scp>UK</scp> Biobank exomes. Genes Chromosomes and Cancer, 2022, 61, 523-529.	2.8	2
98	Integrated in silico and experimental assessment of disease relevance of PCDH19 Âmissense variants. Human Mutation, 2021, 42, 1030-1041.	2.5	1
99	P1â€170: WHOLEâ€EXOME SEQUENCING IN 20,197 INDIVIDUALS IDENTIFIES ULTRAâ€RARE SORL1 LOSSâ€OFâ€FUNCTION VARIANTS IN LATEâ€ONSET ALZHEIMER'S DISEASE. Alzheimer's and Dementia, 2018, 14, P344.	0.8	0