Steven A Mccarroll

List of Publications by Year
in descending order

159 papers	111,990 citations	3531 90 h-index	6300 158 g-index
208 all docs	208 docs citations	208 times ranked	123567 citing authors

#	Article	IF	Citations
1	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	27.8	13,998
2	Analysis of protein-coding genetic variation in 60,706 humans. Nature, 2016, 536, 285-291.	27.8	9,051
3	Finding the missing heritability of complex diseases. Nature, 2009, 461, 747-753.	27.8	7,490
4	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	27.8	6,140
5	Highly Parallel Genome-wide Expression Profiling of Individual Cells Using Nanoliter Droplets. Cell, 2015, 161, 1202-1214.	28.9	5,908
6	Mutational heterogeneity in cancer and the search for new cancer-associated genes. Nature, 2013, 499, 214-218.	27.8	4,761
7	Genetic studies of body mass index yield new insights for obesity biology. Nature, 2015, 518, 197-206.	27.8	3,823
8	Clonal Hematopoiesis and Blood-Cancer Risk Inferred from Blood DNA Sequence. New England Journal of Medicine, 2014, 371, 2477-2487.	27.0	2,669
9	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. Nature Genetics, 2010, 42, 937-948.	21.4	2,634
10	A reference panel of 64,976 haplotypes for genotype imputation. Nature Genetics, 2016, 48, 1279-1283.	21.4	2,421
11	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. Nature Genetics, 2013, 45, 984-994.	21.4	2,067
12	Genes that act downstream of DAF-16 to influence the lifespan of Caenorhabditis elegans. Nature, 2003, 424, 277-283.	27.8	1,998
13	An integrated map of structural variation in 2,504 human genomes. Nature, 2015, 526, 75-81.	27.8	1,994
14	New genetic loci implicated in fasting glucose homeostasis and their impact on type 2 diabetes risk. Nature Genetics, 2010, 42, 105-116.	21.4	1,982
15	Schizophrenia risk from complex variation of complement component 4. Nature, 2016, 530, 177-183.	27.8	1,915
16	Genome-wide detection and characterization of positive selection in human populations. Nature, 2007, 449, 913-918.	27.8	1,788
17	Six new loci associated with body mass index highlight a neuronal influence on body weight regulation. Nature Genetics, 2009, 41, 25-34.	21.4	1,572
18	De novo mutations in schizophrenia implicate synaptic networks. Nature, 2014, 506, 179-184.	27.8	1,510

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19	Genome-wide association analysis identifies 13 new risk loci for schizophrenia. Nature Genetics, 2013, 45, 1150-1159.	21.4	1,395
20	Single-Cell RNA Sequencing of Microglia throughout the Mouse Lifespan and in the Injured Brain Reveals Complex Cell-State Changes. Immunity, 2019, 50, 253-271.e6.	14.3	1,351
21	Common schizophrenia alleles are enriched in mutation-intolerant genes and in regions under strong background selection. Nature Genetics, 2018, 50, 381-389.	21.4	1,332
22	A polygenic burden of rare disruptive mutations in schizophrenia. Nature, 2014, 506, 185-190.	27.8	1,305
23	Molecular Diversity and Specializations among the Cells of the Adult Mouse Brain. Cell, 2018, 174, 1015-1030.e16.	28.9	1,231
24	A Systematic Survey of Loss-of-Function Variants in Human Protein-Coding Genes. Science, 2012, 335, 823-828.	12.6	1,095
25	Comprehensive Classification of Retinal Bipolar Neurons by Single-Cell Transcriptomics. Cell, 2016, 166, 1308-1323.e30.	28.9	1,010
26	Mapping copy number variation by population-scale genome sequencing. Nature, 2011, 470, 59-65.	27.8	991
27	Genome-wide association of early-onset myocardial infarction with single nucleotide polymorphisms and copy number variants. Nature Genetics, 2009, 41, 334-341.	21.4	990
28	Mapping and sequencing of structural variation from eight human genomes. Nature, 2008, 453, 56-64.	27.8	983
29	Cell diversity and network dynamics in photosensitive human brain organoids. Nature, 2017, 545, 48-53.	27.8	933
30	Mapping genomic loci implicates genes and synaptic biology in schizophrenia. Nature, 2022, 604, 502-508.	27.8	929
31	Integrated detection and population-genetic analysis of SNPs and copy number variation. Nature Genetics, 2008, 40, 1166-1174.	21.4	838
32	Contribution of copy number variants to schizophrenia from a genome-wide study of 41,321 subjects. Nature Genetics, 2017, 49, 27-35.	21.4	838
33	Meta-analysis identifies 13 new loci associated with waist-hip ratio and reveals sexual dimorphism in the genetic basis of fat distribution. Nature Genetics, 2010, 42, 949-960.	21.4	836
34	Heritability enrichment of specifically expressed genes identifies disease-relevant tissues and cell types. Nature Genetics, 2018, 50, 621-629.	21.4	807
35	Integrated genotype calling and association analysis of SNPs, common copy number polymorphisms and rare CNVs. Nature Genetics, 2008, 40, 1253-1260.	21.4	712
36	Copy number variation: New insights in genome diversity. Genome Research, 2006, 16, 949-961.	5.5	697

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37	Common deletion polymorphisms in the human genome. Nature Genetics, 2006, 38, 86-92.	21.4	656
38	Multi-platform discovery of haplotype-resolved structural variation in human genomes. Nature Communications, 2019, 10, 1784.	12.8	636
39	A molecular census of arcuate hypothalamus and median eminence cell types. Nature Neuroscience, 2017, 20, 484-496.	14.8	635
40	Genome-wide association study of more than 40,000 bipolar disorder cases provides new insights into the underlying biology. Nature Genetics, 2021, 53, 817-829.	21.4	629
41	Deletion polymorphism upstream of IRGM associated with altered IRGM expression and Crohn's disease. Nature Genetics, 2008, 40, 1107-1112.	21.4	604
42	Partitioning Heritability of Regulatory and Cell-Type-Specific Variants across 11 Common Diseases. American Journal of Human Genetics, 2014, 95, 535-552.	6.2	569
43	Copy-number variation and association studies of human disease. Nature Genetics, 2007, 39, S37-S42.	21.4	531
44	Discovery and Statistical Genotyping of Copy-Number Variation from Whole-Exome Sequencing Depth. American Journal of Human Genetics, 2012, 91, 597-607.	6.2	513
45	Whole-Genome Sequencing in Autism Identifies Hot Spots for De Novo Germline Mutation. Cell, 2012, 151, 1431-1442.	28.9	501
46	Comparative genetic architectures of schizophrenia in East Asian and European populations. Nature Genetics, 2019, 51, 1670-1678.	21.4	440
47	Comparing genomic expression patterns across species identifies shared transcriptional profile in aging. Nature Genetics, 2004, 36, 197-204.	21.4	434
48	Lymphatic and Other Vascular Malformative/Overgrowth Disorders AreÂCaused by Somatic Mutations in PIK3CA. Journal of Pediatrics, 2015, 166, 1048-1054.e5.	1.8	429
49	Increased burden of ultra-rare protein-altering variants among 4,877 individuals with schizophrenia. Nature Neuroscience, 2016, 19, 1433-1441.	14.8	427
50	Human pluripotent stem cells recurrently acquire and expand dominant negative P53 mutations. Nature, 2017, 545, 229-233.	27.8	409
51	Transcriptome-wide association study of schizophrenia and chromatin activity yields mechanistic disease insights. Nature Genetics, 2018, 50, 538-548.	21.4	406
52	Pathways Disrupted in Human ALS Motor Neurons Identified through Genetic Correction of Mutant SOD1. Cell Stem Cell, 2014, 14, 781-795.	11.1	392
53	Analysis of copy number variations at 15 schizophrenia-associated loci. British Journal of Psychiatry, 2014, 204, 108-114.	2.8	380
54	De novo copy number variants identify new genes and loci in isolated sporadic tetralogy of Fallot. Nature Genetics, 2009, 41, 931-935.	21.4	373

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55	Comparative cellular analysis of motor cortex in human, marmoset and mouse. Nature, 2021, 598, 111-119.	27.8	361
56	Large multiallelic copy number variations in humans. Nature Genetics, 2015, 47, 296-303.	21.4	357
57	Rare coding variants in ten genes confer substantial risk for schizophrenia. Nature, 2022, 604, 509-516.	27.8	326
58	The Penetrance of Copy Number Variations for Schizophrenia and Developmental Delay. Biological Psychiatry, 2014, 75, 378-385.	1.3	321
59	A multimodal cell census and atlas of the mammalian primary motor cortex. Nature, 2021, 598, 86-102.	27.8	316
60	Discovery and genotyping of genome structural polymorphism by sequencing on a population scale. Nature Genetics, 2011, 43, 269-276.	21.4	299
61	Increased neutrophil extracellular trap formation promotes thrombosis in myeloproliferative neoplasms. Science Translational Medicine, 2018, 10, .	12.4	299
62	Quantifying prion disease penetrance using large population control cohorts. Science Translational Medicine, 2016, 8, 322ra9.	12.4	289
63	Linkage Disequilibrium and Heritability of Copy-Number Polymorphisms within Duplicated Regions of the Human Genome. American Journal of Human Genetics, 2006, 79, 275-290.	6.2	283
64	Identification of heart rateâ€“associated loci and their effects on cardiac conduction and rhythm disorders. Nature Genetics, 2013, 45, 621-631.	21.4	282
65	Insights into clonal haematopoiesis from 8,342 mosaic chromosomal alterations. Nature, 2018, 559, 350-355.	27.8	279
66	Punctuated bursts in human male demography inferred from 1,244 worldwide Y-chromosome sequences. Nature Genetics, 2016, 48, 593-599.	21.4	273
67	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
68	An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. Nature Genetics, 2018, 50, 727-736.	21.4	235
69	Differential Relationship of DNA Replication Timing to Different Forms of Human Mutation and Variation. American Journal of Human Genetics, 2012, 91, 1033-1040.	6.2	220
70	Single-cell RNA sequencing reveals compromised immune microenvironment in precursor stages of multiple myeloma. Nature Cancer, 2020, 1, 493-506.	13.2	209
71	Innovations present in the primate interneuron repertoire. Nature, 2020, 586, 262-269.	27.8	206
72	Genetic predisposition to mosaic Y chromosome loss in blood. Nature, 2019, 575, 652-657.	27.8	198

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73	zCall: a rare variant caller for array-based genotyping. Bioinformatics, 2012, 28, 2543-2545.	4.1	195
74	The Genetic Landscape of Diamond-Blackfan Anemia. American Journal of Human Genetics, 2018, 103, 930-947.	6.2	184
75	Evaluation of Common Variants in the Six Known Maturity-Onset Diabetes of the Young (MODY) Genes for Association With Type 2 Diabetes. Diabetes, 2007, 56, 685-693.	0.6	178
76	Single-Tissue and Cross-Tissue Heritability of Gene Expression Via Identity-by-Descent in Related or Unrelated Individuals. PLoS Genetics, 2011, 7, e1001317.	3.5	173
77	Novel Findings from CNVs Implicate Inhibitory and Excitatory Signaling Complexes in Schizophrenia. Neuron, 2015, 86, 1203-1214.	8.1	173
78	Phenotypic Landscape of Schizophrenia-Associated Genes Defines Candidates and Their Shared Functions. Cell, 2019, 177, 478-491.e20.	28.9	159
79	Complement genes contribute sex-biased vulnerability in diverse disorders. Nature, 2020, 582, 577-581.	27.8	158
80	Overexpression of schizophrenia susceptibility factor human complement C4A promotes excessive synaptic loss and behavioral changes in mice. Nature Neuroscience, 2021, 24, 214-224.	14.8	158
81	Extending genome-wide association studies to copy-number variation. Human Molecular Genetics, 2008, 17, R135-R142.	2.9	154
82	Genetic Variation in Human DNA Replication Timing. Cell, 2014, 159, 1015-1026.	28.9	149
83	Genetically Distinct Parallel Pathways in the Entopeduncular Nucleus for Limbic and Sensorimotor Output of the Basal Ganglia. Neuron, 2017, 94, 138-152.e5.	8.1	146
84	Accurately Assessing the Risk of Schizophrenia Conferred by Rare Copy-Number Variation Affecting Genes with Brain Function. PLoS Genetics, 2010, 6, e1001097.	3.5	134
85	DNA sequence of human chromosome 17 and analysis of rearrangement in the human lineage. Nature, 2006, 440, 1045-1049.	27.8	130
86	Exome sequencing in schizophrenia-affected parentâ€“offspring trios reveals risk conferred by protein-coding de novo mutations. Nature Neuroscience, 2020, 23, 185-193.	14.8	125
87	Structural haplotypes and recent evolution of the human 17q21.31 region. Nature Genetics, 2012, 44, 881-885.	21.4	124
88	Whole genome sequencing in psychiatric disorders: the WGSPD consortium. Nature Neuroscience, 2017, 20, 1661-1668.	14.8	122
89	Structural forms of the human amylase locus and their relationships to SNPs, haplotypes and obesity. Nature Genetics, 2015, 47, 921-925.	21.4	120
90	Monogenic and polygenic inheritance become instruments for clonal selection. Nature, 2020, 584, 136-141.	27.8	119

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91	Hematopoietic mosaic chromosomal alterations increase the risk for diverse types of infection. Nature Medicine, 2021, 27, 1012-1024.	30.7	109
92	Evidence of Common Genetic Overlap Between Schizophrenia and Cognition. Schizophrenia Bulletin, 2016, 42, 832-842.	4.3	102
93	Chromosomal alterations among age-related haematopoietic clones in Japan. Nature, 2020, 584, 130-135.	27.8	102
94	Protein-coding repeat polymorphisms strongly shape diverse human phenotypes. Science, 2021, 373, 1499-1505.	12.6	96
95	Recurring exon deletions in the HP (haptoglobin) gene contribute to lower blood cholesterol levels. Nature Genetics, 2016, 48, 359-366.	21.4	93
96	Donor-recipient mismatch for common gene deletion polymorphisms in graft-versus-host disease. Nature Genetics, 2009, 41, 1341-1344.	21.4	91
97	Ultra-rare disruptive and damaging mutations influence educational attainment in the general population. Nature Neuroscience, 2016, 19, 1563-1565.	14.8	90
98	CNV analysis in a large schizophrenia sample implicates deletions at 16p12.1 and SLC1A1 and duplications at 1p36.33 and CGNL1. Human Molecular Genetics, 2014, 23, 1669-1676.	2.9	82
99	Genome-scale neurogenetics: methodology and meaning. Nature Neuroscience, 2014, 17, 756-763.	14.8	82
100	Contributions of common genetic variants to risk of schizophrenia among individuals of African and Latino ancestry. Molecular Psychiatry, 2020, 25, 2455-2467.	7.9	82
101	Genetic association analysis highlights new loci that modulate hematological trait variation in Caucasians and African Americans. Human Genetics, 2011, 129, 307-317.	3.8	81
102	Deep coverage whole genome sequences and plasma lipoprotein(a) in individuals of European and African ancestries. Nature Communications, 2018, 9, 2606.	12.8	79
103	Progress in the Genetics of Polygenic Brain Disorders: Significant New Challenges for Neurobiology. Neuron, 2013, 80, 578-587.	8.1	74
104	A whole-genome sequence study identifies genetic risk factors for neuromyelitis optica. Nature Communications, 2018, 9, 1929.	12.8	73
105	Insights into variation in meiosis from 31,228 human sperm genomes. Nature, 2020, 583, 259-264.	27.8	73
106	De novo CNVs in bipolar affective disorder and schizophrenia. Human Molecular Genetics, 2014, 23, 6677-6683.	2.9	70
107	Genome-wide association study identifies SESTD1 as a novel risk gene for lithium-responsive bipolar disorder. Molecular Psychiatry, 2016, 21, 1290-1297.	7.9	69
108	Random replication of the inactive X chromosome. Genome Research, 2014, 24, 64-69.	5.5	65

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109	Mapping a New Spontaneous Preterm Birth Susceptibility Gene, IGF1R, Using Linkage, Haplotype Sharing, and Association Analysis. PLoS Genetics, 2011, 7, e1001293.	3.5	61
110	Using population admixture to help complete maps of the human genome. Nature Genetics, 2013, 45, 406-414.	21.4	61
111	A Rapid Molecular Approach for Chromosomal Phasing. PLoS ONE, 2015, 10, e0118270.	2.5	58
112	Genome-wide association analysis of red blood cell traits in African Americans: the COGENT Network. Human Molecular Genetics, 2013, 22, 2529-2538.	2.9	57
113	Clonal Hematopoiesis and Blood-Cancer Risk. New England Journal of Medicine, 2015, 372, 1071-1072.	27.0	57
114	SnapShot-Seq: A Method for Extracting Genome-Wide, In Vivo mRNA Dynamics from a Single Total RNA Sample. PLoS ONE, 2014, 9, e89673.	2.5	53
115	New data and an old puzzle: the negative association between schizophrenia and rheumatoid arthritis. International Journal of Epidemiology, 2015, 44, 1706-1721.	1.9	53
116	A Loss-of-Function Splice Acceptor Variant in IGF2 Is Protective for Type 2 Diabetes. Diabetes, 2017, 66, 2903-2914.	0.6	52
117	Thy-1 Is a Component Common to Multiple Populations of Synaptic Vesicles. Journal of Cell Biology, 1998, 140, 685-698.	5.2	51
118	Complex and multi-allelic copy number variation in human disease. Briefings in Functional Genomics, 2015, 14, 329-338.	2.7	50
119	Polygenic risk for schizophrenia and measured domains of cognition in individuals with psychosis and controls. Translational Psychiatry, 2018, 8, 78.	4.8	49
120	Genome-Wide Association Studies of Schizophrenia and Bipolar Disorder in a Diverse Cohort of US Veterans. Schizophrenia Bulletin, 2021, 47, 517-529.	4.3	48
121	Cryptic and Complex Chromosomal Aberrations in Early-Onset Neuropsychiatric Disorders. American Journal of Human Genetics, 2014, 95, 454-461.	6.2	45
122	Common Î±-globin variants modify hematologic and other clinical phenotypes in sickle cell trait and disease. PLoS Genetics, 2018, 14, e1007293.	3.5	45
123	Contribution of Rare Copy Number Variants toÂBipolar Disorder Risk Is Limited to Schizoaffective Cases. Biological Psychiatry, 2019, 86, 110-119.	1.3	45
124	Identification of Transcriptional Regulatory Elements in Chemosensory Receptor Genes by Probabilistic Segmentation. Current Biology, 2005, 15, 347-352.	3.9	42
125	Analyzing Copy Number Variation with Droplet Digital PCR. Methods in Molecular Biology, 2018, 1768, 143-160.	0.9	37
126	Mapping the Human Reference Genomeâ€™s Missing Sequence by Three-Way Admixture in Latino Genomes. American Journal of Human Genetics, 2013, 93, 411-421.	6.2	36

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127	Copy-number analysis goes more than skin deep. Nature Genetics, 2008, 40, 5-6.	21.4	35
128	Anterior thalamic dysfunction underlies cognitive deficits in a subset of neuropsychiatric disease models. Neuron, 2021, 109, 2590-2603.e13.	8.1	34
129	Prognostic value of polygenic risk scores for adults with psychosis. Nature Medicine, 2021, 27, 1576-1581.	30.7	31
130	Improved detection of global copy number variation using high density, non-polymorphic oligonucleotide probes. BMC Genetics, 2008, 9, 27.	2.7	27
131	Whole-genome analysis of human embryonic stem cells enables rational line selection based on genetic variation. Cell Stem Cell, 2022, 29, 472-486.e7.	11.1	27
132	Polygenic risk for type 2 diabetes mellitus among individuals with psychosis and their relatives. Journal of Psychiatric Research, 2016, 77, 52-58.	3.1	22
133	The 22q11.2 region regulates presynaptic gene-products linked to schizophrenia. Nature Communications, 2022, 13, .	12.8	22
134	Exploring the variation within. Nature Genetics, 2012, 44, 614-616.	21.4	21
135	No evidence for rare recessive and compound heterozygous disruptive variants in schizophrenia. European Journal of Human Genetics, 2015, 23, 555-557.	2.8	21
136	Human DDK rescues stalled forks and counteracts checkpoint inhibition at unfired origins to complete DNA replication. Molecular Cell, 2021, 81, 426-441.e8.	9.7	21
137	Copy number variation and human genome maps. Nature Genetics, 2010, 42, 365-366.	21.4	20
138	Early role for a Na ⁺ ,K ⁺ -ATPase (ATP1A3) in brain development. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	20
139	Mutation intolerant genes and targets of FMRP are enriched for nonsynonymous alleles in schizophrenia. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2017, 174, 724-731.	1.7	19
140	Differential excitatory vs inhibitory SCN expression at single cell level regulates brain sodium channel function in neurodevelopmental disorders. European Journal of Paediatric Neurology, 2020, 24, 129-133.	1.6	18
141	Publicly Available hiPSC Lines with Extreme Polygenic Risk Scores for Modeling Schizophrenia. Complex Psychiatry, 2020, 6, 68-82.	0.9	18
142	Investigating rare pathogenic/likely pathogenic exonic variation in bipolar disorder. Molecular Psychiatry, 2021, 26, 5239-5250.	7.9	15
143	New insights into the biological basis of genomic disorders. Nature Genetics, 2006, 38, 1363-1364.	21.4	12
144	On the level: IRGM gene function is all about expression. Autophagy, 2009, 5, 96-99.	9.1	10

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145	Our Fallen Genomes. Science, 2013, 342, 564-565.	12.6	8
146	Using Droplet Digital PCR to Analyze Allele-Specific RNA Expression. Methods in Molecular Biology, 2018, 1768, 401-422.	0.9	8
147	Mapping duplicated sequences. Nature Biotechnology, 2009, 27, 1001-1002.	17.5	5
148	Of Rats and Men. Cell, 2013, 154, 481-483.	28.9	4
149	Single cell analysis of DNA in more than 10,000 individual sperm from men with abnormal reproductive outcomes. Journal of Assisted Reproduction and Genetics, 2021, 38, 2975-2983.	2.5	2
150	Single-Cell RNA Sequencing Reveals Compromised Immune Microenvironment in Precursor Stages of Multiple Myeloma. Blood, 2018, 132, 2603-2603.	1.4	1
151	Chromosomal phase improves aneuploidy detection in non-invasive prenatal testing at low fetal DNA fractions. Scientific Reports, 2022, 12, .	3.3	1
152	Population Perspectives on Genome Variation and Complex Disease. , 2013, , 41-49.		0
153	Damaging Missense De Novo Coding Mutations Contribute To Schizophrenia Risk. European Neuropsychopharmacology, 2017, 27, S427-S428.	0.7	0
154	Ultra-Rare Protein-Altering Variants Among 4,877 Swedish Individuals with Schizophrenia. European Neuropsychopharmacology, 2017, 27, S426-S427.	0.7	0
155	SA122STRUCTURAL VARIATIONS OF SCHIZOPHRENIA RISK GENE COMPLEMENT COMPONENT 4 (C4) AND BRAIN MRI PHENOTYPES. European Neuropsychopharmacology, 2019, 29, S1255-S1256.	0.7	0
156	THE GENOMICS OF BIPOLAR AND SCHIZOPHRENIC DISORDERS IN A LARGE PEDIGREE FROM A NORTHERN SWEDISH ISOLATE. European Neuropsychopharmacology, 2019, 29, S902-S903.	0.7	0
157	169. New Technology for Learning About Genetic EffectsÂon Brain Cells and Brain Tissue. Biological Psychiatry, 2019, 85, S70.	1.3	0
158	Single-cell RNA sequencing reveals compromised immune microenvironment in precursor stages of multiple myeloma. Clinical Lymphoma, Myeloma and Leukemia, 2019, 19, e27.	0.4	0
159	Genome Variation and Donor-Recipient Compatibility in Graft-Versus-Host Disease. Blood, 2010, 116, SCI-12-SCI-12.	1.4	0