Michael E Talkowski

List of Publications by Year
in descending order

122 papers	20,162 citations	53939 47 h-index	20625 120 g-index
167 all docs	167 docs citations	167 times ranked	36271 citing authors

#	Article	IF	Citations
1	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	13.7	6,140
2	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. Cell, 2020, 180, 568-584.e23.	13.5	1,422
3	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. Neuron, 2015, 87, 1215-1233.	3.8	1,219
4	Multi-platform discovery of haplotype-resolved structural variation in human genomes. Nature Communications, 2019, 10, 1784.	5.8	636
5	A structural variation reference for medical and population genetics. Nature, 2020, 581, 444-451.	13.7	614
6	Sequencing Chromosomal Abnormalities Reveals Neurodevelopmental Loci that Confer Risk across Diagnostic Boundaries. Cell, 2012, 149, 525-537.	13.5	534
7	Low Incidence of Off-Target Mutations in Individual CRISPR-Cas9 and TALEN Targeted Human Stem Cell Clones Detected by Whole-Genome Sequencing. Cell Stem Cell, 2014, 15, 27-30.	5.2	456
8	Efficient Ablation of Genes in Human Hematopoietic Stem and Effector Cells using CRISPR/Cas9. Cell Stem Cell, 2014, 15, 643-652.	5.2	406
9	KCTD13 is a major driver of mirrored neuroanatomical phenotypes of the 16p11.2 copy number variant. Nature, 2012, 485, 363-367.	13.7	363
10	Haplotype-resolved diverse human genomes and integrated analysis of structural variation. Science, 2021, 372, .	6.0	358
11	Mapping and phasing of structural variation in patient genomes using nanopore sequencing. Nature Communications, 2017, 8, 1326.	5.8	315
12	CHD8 regulates neurodevelopmental pathways associated with autism spectrum disorder in neural progenitors. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, E4468-77.	3.3	297
13	Insights into clonal haematopoiesis from 8,342 mosaic chromosomal alterations. Nature, 2018, 559, 350-355.	13.7	279
14	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45.	9.4	251
15	An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. Nature Genetics, 2018, 50, 727-736.	9.4	235
16	Genome-wide de novo risk score implicates promoter variation in autism spectrum disorder. Science, 2018, 362, .	6.0	234
17	Complex reorganization and predominant non-homologous repair following chromosomal breakage in karyotypically balanced germline rearrangements and transgenic integration. Nature Genetics, 2012, 44, 390-397.	9.4	229
18	Whole-Genome Sequencing to Characterize Monogenic and Polygenic Contributions in Patients Hospitalized With Early-Onset Myocardial Infarction. Circulation, 2019, 139, 1593-1602.	1.6	213

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19	Assessment of 2q23.1 Microdeletion Syndrome Implicates MBD5 as a Single Causal Locus of Intellectual Disability, Epilepsy, and Autism Spectrum Disorder. American Journal of Human Genetics, 2011, 89, 551-563.	2.6	195
20	An Ancient, Unified Mechanism for Metformin Growth Inhibition in C.Âelegans and Cancer. Cell, 2016, 167, 1705-1718.e13.	13.5	181
21	Clinical Diagnosis by Whole-Genome Sequencing of a Prenatal Sample. New England Journal of Medicine, 2012, 367, 2226-2232.	13.9	174
22	Dissecting the Causal Mechanism of X-Linked Dystonia-Parkinsonism by Integrating Genome and Transcriptome Assembly. Cell, 2018, 172, 897-909.e21.	13.5	163
23	Defining the diverse spectrum of inversions, complex structural variation, and chromothripsis in the morbid humanÂgenome. Genome Biology, 2017, 18, 36.	3.8	159
24	Next-Generation Sequencing Strategies Enable Routine Detection of Balanced Chromosome Rearrangements for Clinical Diagnostics and Genetic Research. American Journal of Human Genetics, 2011, 88, 469-481.	2.6	154
25	Mutations in DCHS1 cause mitral valve prolapse. Nature, 2015, 525, 109-113.	13.7	150
26	Loss of Î´-catenin function in severe autism. Nature, 2015, 520, 51-56.	13.7	145
27	Disruption of the ASTN2/TRIM32 locus at 9q33.1 is a risk factor in males for autism spectrum disorders, ADHD and other neurodevelopmental phenotypes. Human Molecular Genetics, 2014, 23, 2752-2768.	1.4	140
28	Exonic Deletions in AUTS2 Cause a Syndromic Form of Intellectual Disability and Suggest a Critical Role for the C Terminus. American Journal of Human Genetics, 2013, 92, 210-220.	2.6	135
29	Association study of 21 circadian genes with bipolar I disorder, schizoaffective disorder, and schizophrenia. Bipolar Disorders, 2009, 11, 701-710.	1.1	133
30	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248.	9.4	131
31	A network of dopaminergic gene variations implicated as risk factors for schizophrenia. Human Molecular Genetics, 2008, 17, 747-758.	1.4	124
32	Transcriptional Consequences of 16p11.2 Deletion and Duplication in Mouse Cortex and Multiplex Autism Families. American Journal of Human Genetics, 2014, 94, 870-883.	2.6	116
33	Cognitive influences in postural control of patients with unilateral vestibular loss. Gait and Posture, 2004, 19, 105-114.	0.6	113
34	Whole-Genome and RNA Sequencing Reveal Variation and Transcriptomic Coordination in the Developing Human Prefrontal Cortex. Cell Reports, 2020, 31, 107489.	2.9	91
35	Evaluation of a Susceptibility Gene for Schizophrenia: Genotype Based Meta-Analysis of RGS4 Polymorphisms from Thirteen Independent Samples. Biological Psychiatry, 2006, 60, 152-162.	0.7	87
36	Haploinsufficiency ofSOX5at 12p12.1 is associated with developmental delays with prominent language delay, behavior problems, and mild dysmorphic features. Human Mutation, 2012, 33, 728-740.	1.1	85

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37	Ectopic expression of RAD52 and dn53BP1 improves homology-directed repair during CRISPRâ€“Cas9 genome editing. Nature Biomedical Engineering, 2017, 1, 878-888.	11.6	83
38	Primary cilia defects causing mitral valve prolapse. Science Translational Medicine, 2019, 11, .	5.8	76
39	Engineering microdeletions and microduplications by targeting segmental duplications with CRISPR. Nature Neuroscience, 2016, 19, 517-522.	7.1	72
40	Expectations and blind spots for structural variation detection from long-read assemblies and short-read genome sequencing technologies. American Journal of Human Genetics, 2021, 108, 919-928.	2.6	72
41	SYCP2 Translocation-Mediated Dysregulation and Frameshift Variants Cause Human Male Infertility. American Journal of Human Genetics, 2020, 106, 41-57.	2.6	66
42	Novel, Replicated Associations Between Dopamine D3 Receptor Gene Polymorphisms and Schizophrenia in Two Independent Samples. Biological Psychiatry, 2006, 60, 570-577.	0.7	62
43	Disruption of a Large Intergenic Noncoding RNA in Subjects with Neurodevelopmental Disabilities. American Journal of Human Genetics, 2012, 91, 1128-1134.	2.6	61
44	Haploinsufficiency of KDM6A is associated with severe psychomotor retardation, global growth restriction, seizures and cleft palate. Human Genetics, 2013, 132, 537-552.	1.8	60
45	Translocations Disrupting PHF21A in the Potocki-Shaffer-Syndrome Region Are Associated with Intellectual Disability and Craniofacial Anomalies. American Journal of Human Genetics, 2012, 91, 56-72.	2.6	59
46	Autism Spectrum Disorder Genetics. Harvard Review of Psychiatry, 2014, 22, 65-75.	0.9	59
47	De novo structural mutation rates and gamete-of-origin biases revealed through genome sequencing of 2,396 families. American Journal of Human Genetics, 2021, 108, 597-607.	2.6	57
48	Htt CAG repeat expansion confers pleiotropic gains of mutant huntingtin function in chromatin regulation. Human Molecular Genetics, 2015, 24, 2442-2457.	1.4	53
49	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. American Journal of Human Genetics, 2015, 96, 784-796.	2.6	53
50	Structural Chromosomal Rearrangements Require Nucleotide-Level Resolution: Lessons from Next-Generation Sequencing in Prenatal Diagnosis. American Journal of Human Genetics, 2016, 99, 1015-1033.	2.6	53
51	Computational Prediction of Position Effects of Apparently Balanced Human Chromosomal Rearrangements. American Journal of Human Genetics, 2017, 101, 206-217.	2.6	51
52	The cell adhesion gene PVRL3 is associated with congenital ocular defects. Human Genetics, 2012, 131, 235-250.	1.8	46
53	Cryptic and Complex Chromosomal Aberrations in Early-Onset Neuropsychiatric Disorders. American Journal of Human Genetics, 2014, 95, 454-461.	2.6	45
54	Paired-Duplication Signatures Mark Cryptic Inversions and Other Complex Structural Variation. American Journal of Human Genetics, 2015, 97, 170-176.	2.6	45

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55	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4.	13.7	45
56	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	1.1	44
57	Molecular Analysis of a Deletion Hotspot in the NRXN1 Region Reveals the Involvement of Short Inverted Repeats in Deletion CNVs. American Journal of Human Genetics, 2013, 92, 375-386.	2.6	42
58	Describing Sequencing Results of Structural Chromosome Rearrangements with a Suggested Next-Generation Cytogenetic Nomenclature. American Journal of Human Genetics, 2014, 94, 695-709.	2.6	42
59	Reciprocal deletion and duplication at 2q23.1 indicates a role for MBD5 in autism spectrum disorder. European Journal of Human Genetics, 2014, 22, 57-63.	1.4	42
60	Implication of LRRC4C and DPP6 in neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2017, 173, 395-406.	0.7	40
61	Loss of MAGEL2 in Prader-Willi syndrome leads to decreased secretory granule and neuropeptide production. JCI Insight, 2020, 5, .	2.3	40
62	Serotonin gene polymorphisms and bipolar I disorder: Focus on the serotonin transporter. Annals of Medicine, 2005, 37, 590-602.	1.5	39
63	Dopamine Genes and Schizophrenia: Case Closed or Evidence Pending?. Schizophrenia Bulletin, 2007, 33, 1071-1081.	2.3	37
64	Indexcov: fast coverage quality control for whole-genome sequencing. GigaScience, 2017, 6, 1-6.	3.3	36
65	16p11.2 deletion is associated with hyperactivation of human iPSC-derived dopaminergic neuron networks and is rescued by RHOA inhibition in vitro. Nature Communications, 2021, 12, 2897.	5.8	35
66	Linkage Disequilibrium Patterns and Functional Analysis of RGS4 Polymorphisms in Relation to Schizophrenia. Schizophrenia Bulletin, 2007, 34, 118-126.	2.3	34
67	Kctd13-deficient mice display short-term memory impairment and sex-dependent genetic interactions. Human Molecular Genetics, 2019, 28, 1474-1486.	1.4	32
68	Functional annotation of rare structural variation in the human brain. Nature Communications, 2020, 11, 2990.	5.8	32
69	An eMERGE Clinical Center at Partners Personalized Medicine. Journal of Personalized Medicine, 2016, 6, 5.	1.1	31
70	Actin capping protein CAPZB regulates cell morphology, differentiation, and neural crest migration in craniofacial morphogenesis. Human Molecular Genetics, 2016, 25, 1255-1270.	1.4	30
71	The female protective effect against autism spectrum disorder. Cell Genomics, 2022, 2, 100134.	3.0	30
72	Mechanisms for Structural Variation in the Human Genome. Current Genetic Medicine Reports, 2013, 1, 81-90.	1.9	29

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73	Risks and Recommendations in Prenatally Detected De Novo Balanced Chromosomal Rearrangements from Assessment of Long-Term Outcomes. American Journal of Human Genetics, 2018, 102, 1090-1103.	2.6	29
74	TSC patient-derived isogenic neural progenitor cells reveal altered early neurodevelopmental phenotypes and rapamycin-induced MNK-eIF4E signaling. Molecular Autism, 2020, 11, 2.	2.6	29
75	Consanguinity associated with increased risk for bipolar I disorder in Egypt. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2009, 150B, 879-885.	1.1	28
76	A Balanced Translocation in Kallmann Syndrome Implicates a Long Noncoding RNA, RMST, as a GnRH Neuronal Regulator. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e231-e244.	1.8	28
77	New gene discoveries highlight functional convergence in autism and related neurodevelopmental disorders. Current Opinion in Genetics and Development, 2020, 65, 195-206.	1.5	27
78	Genome-encoded cytoplasmic double-stranded RNAs, found in C9ORF72 ALS-FTD brain, propagate neuronal loss. Science Translational Medicine, 2021, 13, .	5.8	27
79	Whole exome sequencing analyses reveal geneâ€“microbiota interactions in the context of IBD. Gut, 2021, 70, gutjnl-2019-319706.	6.1	26
80	A deep learning approach to identify gene targets of a therapeutic for human splicing disorders. Nature Communications, 2021, 12, 3332.	5.8	26
81	Genetic associations between neuregulin-1 SNPs and neurocognitive function in multigenerational, multiplex schizophrenia families. Psychiatric Genetics, 2012, 22, 70-81.	0.6	23
82	Highly Penetrant Alterations of a Critical Region Including BDNF in Human Psychopathology and Obesity. Archives of General Psychiatry, 2012, 69, 1238.	13.8	22
83	Mutated Huntingtin Causes Testicular Pathology in Transgenic Minipig Boars. Neurodegenerative Diseases, 2016, 16, 245-259.	0.8	22
84	Evidence for secondary-variant genetic burden and non-random distribution across biological modules in a recessive ciliopathy. Nature Genetics, 2020, 52, 1145-1150.	9.4	22
85	Prevalence and phenotypic impact of rare potentially damaging variants in autism spectrum disorder. Molecular Autism, 2021, 12, 65.	2.6	22
86	A comprehensive genetic association and functional study of TNF in schizophrenia risk. Schizophrenia Research, 2006, 83, 7-13.	1.1	21
87	Genomic and Functional Overlap between Somatic and Germline Chromosomal Rearrangements. Cell Reports, 2014, 9, 2001-2010.	2.9	21
88	Familial thrombocytopenia due to a complex structural variant resulting in a WAC-ANKRD26 fusion transcript. Journal of Experimental Medicine, 2021, 218, .	4.2	20
89	Can RGS4 Polymorphisms Be Viewed as Credible Risk Factors for Schizophrenia? A Critical Review of the Evidence. Schizophrenia Bulletin, 2006, 32, 203-208.	2.3	19
90	WNT/Î²-Catenin Pathway and Epigenetic Mechanisms Regulate the Pitt-Hopkins Syndrome and Schizophrenia Risk Gene TCF4. Molecular Neuropsychiatry, 2017, 3, 53-71.	3.0	19

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91	Prioritization of genes driving congenital phenotypes of patients with de novo genomic structural variants. Genome Medicine, 2019, 11, 79.	3.6	19
92	Systematic Association Studies of Mitochondrial DNA Variations in Schizophrenia: Focus on the ND5 Gene. Schizophrenia Bulletin, 2008, 34, 458-465.	2.3	18
93	Fineâ€mapping reveals novel alternative splicing of the dopamine transporter. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 1434-1447.	1.1	18
94	RGS4 Polymorphisms Associated With Variability of Cognitive Performance in a Family-Based Schizophrenia Sample. Schizophrenia Bulletin, 2010, 36, 983-990.	2.3	18
95	Lack of association of rare functional variants in TSC1/TSC2 genes with autism spectrum disorder. Molecular Autism, 2013, 4, 5.	2.6	16
96	Convergent patterns of association between phenylalanine hydroxylase variants and schizophrenia in four independent samples. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2009, 150B, 560-569.	1.1	15
97	Mosaic copy number variation in schizophrenia. European Journal of Human Genetics, 2013, 21, 1007-1011.	1.4	15
98	Potential molecular consequences of transgene integration: The R6/2 mouse example. Scientific Reports, 2017, 7, 41120.	1.6	14
99	Introduction of genomics into prenatal diagnostics. Lancet, The, 2019, 393, 719-721.	6.3	13
100	Hypomorphic mutation of the mouse Huntingtonâ€™s disease gene orthologue. PLoS Genetics, 2019, 15, e1007765.	1.5	13
101	Biallelic mutation of FBXL7 suggests a novel form of Hennekam syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 189-194.	0.7	13
102	Dystonia-specific mutations in THAP1 alter transcription of genes associated with neurodevelopment and myelin. American Journal of Human Genetics, 2021, 108, 2145-2158.	2.6	13
103	Estrogen-related receptor gamma implicated in a phenotype including hearing loss and mild developmental delay. European Journal of Human Genetics, 2016, 24, 1622-1626.	1.4	12
104	Transcriptional consequences of MBD5 disruption in mouse brain and CRISPR-derived neurons. Molecular Autism, 2020, 11, 45.	2.6	11
105	Orgo-Seq integrates single-cell and bulk transcriptomic data to identify cell type specific-driver genes associated with autism spectrum disorder. Nature Communications, 2022, 13, .	5.8	11
106	Functional Analysis of Upstream Common Polymorphisms of the Dopamine Transporter Gene. Schizophrenia Bulletin, 2010, 36, 977-982.	2.3	10
107	Prevalence and Phenotypic Effects of Copy Number Variants in Isolated Hypogonadotropic Hypogonadism. Journal of Clinical Endocrinology and Metabolism, 2022, 107, 2228-2242.	1.8	10
108	Histone deacetylase knockouts modify transcription, CAG instability and nuclear pathology in Huntington disease mice. ELife, 2020, 9, .	2.8	9

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109	Phenotypic interpretation of complex chromosomal rearrangements informed by nucleotide-level resolution and structural organization of chromatin. European Journal of Human Genetics, 2018, 26, 374-381.	1.4	8
110	A neurodevelopmental disorder caused by a novel de novo SVA insertion in exon 13 of the SRCAP gene. European Journal of Human Genetics, 2022, 30, 1083-1087.	1.4	8
111	Complex and Dynamic Chromosomal Rearrangements in a Family With Seemingly Non-Mendelian Inheritance of Dopa-Responsive Dystonia. JAMA Neurology, 2017, 74, 806.	4.5	7
112	A novel microduplication of ARID1B: Clinical, genetic, and proteomic findings. American Journal of Medical Genetics, Part A, 2017, 173, 2478-2484.	0.7	7
113	Developmental regulation of neuronal gene expression by Elongator complex protein 1 dosage. Journal of Genetics and Genomics, 2022, 49, 654-665.	1.7	6
114	Role of the Chromosome Architectural Factor SMCHD1 in X-Chromosome Inactivation, Gene Regulation, and Disease in Humans. Genetics, 2019, 213, 685-703.	1.2	5
115	The Role of Attention in Vestibular Processing. Proceedings of the Human Factors and Ergonomics Society, 2002, 46, 255-259.	0.2	1
116	Age dependent association of inbreeding with risk for schizophrenia in Egypt. Schizophrenia Research, 2020, 216, 450-459.	1.1	1
117	Xenopus models suggest convergence of gene signatures on neurogenesis in autism. Neuron, 2021, 109, 743-745.	3.8	1
118	Cover Image, Volume 173A, Number 2, February 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	0.7	0
119	20.1 DISSECTING THE FUNCTIONAL CONSEQUENCES OF RECIPROCAL GENOMIC DISORDERS. Schizophrenia Bulletin, 2018, 44, S33-S33.	2.3	0
120	Next Generation Sequencing of Prenatal Structural Chromosomal Rearrangements Using Large-Insert Libraries. Methods in Molecular Biology, 2019, 1885, 251-265.	0.4	0
121	Contribution of Copy Number Variation in Idiopathic Hypogonadotropic Hypogonadism. Journal of the Endocrine Society, 2021, 5, A756-A756.	0.1	0
122	Dissecting the Causal Mechanism of X-Linked Dystonia-Parkinsonism by Integrating Genome and Transcriptome Assembly. SSRN Electronic Journal, 0, , .	0.4	0