Michael E Talkowski

List of Publications by Citations

135 papers	11,186 citations	43 h-index	105 g-index
167 ext. papers	16,717 ext. citations	14.1 avg, IF	5.58 L-index

#	Paper	IF	Citations
135	The mutational constraint spectrum quantified from variation in 141,456 humans. *Nature*, **2020**, 581, 434-443	50.4	2278
134	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. *Neuron*, **2015**, 87, 1215-1233	13.9	806
133	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. *Cell*, **2020**, 180, 568-584.e23	56.2	578
132	Sequencing chromosomal abnormalities reveals neurodevelopmental loci that confer risk across diagnostic boundaries. *Cell*, **2012**, 149, 525-37	56.2	441
131	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	18	394
130	The mutational constraint spectrum quantified from variation in 141,456 humans		381
129	Multi-platform discovery of haplotype-resolved structural variation in human genomes. *Nature Communications*, **2019**, 10, 1784	17.4	346
128	Efficient ablation of genes in human hematopoietic stem and effector cells using CRISPR/Cas9. *Cell Stem Cell*, **2014**, 15, 643-52	18	324
127	KCTD13 is a major driver of mirrored neuroanatomical phenotypes of the 16p11.2 copy number variant. *Nature*, **2012**, 485, 363-7	50.4	281
126	A structural variation reference for medical and population genetics. *Nature*, **2020**, 581, 444-451	50.4	223
125	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	11.5	210
124	Mapping and phasing of structural variation in patient genomes using nanopore sequencing. *Nature Communications*, **2017**, 8, 1326	17.4	191
123	Complex reorganization and predominant non-homologous repair following chromosomal breakage in karyotypically balanced germline rearrangements and transgenic integration. *Nature Genetics*, **2012**, 44, 390-7, S1	36.3	190
122	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. *Nature Genetics*, **2017**, 49, 36-45	36.3	172
121	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, 551-63	11	166
120	An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. *Nature Genetics*, **2018**, 50, 727-736	36.3	156
119	Insights into clonal haematopoiesis from 8,342 mosaic chromosomal alterations. *Nature*, **2018**, 559, 350-355	36.4	144

118	Clinical diagnosis by whole-genome sequencing of a prenatal sample. *New England Journal of Medicine*, **2012**, 367, 2226-32	59.2	144
117	An Ancient, Unified Mechanism for Metformin Growth Inhibition in C. elegans and Cancer. *Cell*, **2016**, 167, 1705-1718.e13	56.2	134
116	Genome-wide de novo risk score implicates promoter variation in autism spectrum disorder. *Science* , **2018**, 362,	33.3	134
115	Next-generation sequencing strategies enable routine detection of balanced chromosome rearrangements for clinical diagnostics and genetic research. *American Journal of Human Genetics*, **2011**, 88, 469-81	11	132
114	Association study of 21 circadian genes with bipolar I disorder, schizoaffective disorder, and schizophrenia. *Bipolar Disorders*, **2009**, 11, 701-10	3.8	120
113	A network of dopaminergic gene variations implicated as risk factors for schizophrenia. *Human Molecular Genetics*, **2008**, 17, 747-58	5.6	120
112	Whole-Genome Sequencing to Characterize Monogenic and Polygenic Contributions in Patients Hospitalized With Early-Onset Myocardial Infarction. *Circulation*, **2019**, 139, 1593-1602	16.7	112
111	Defining the diverse spectrum of inversions, complex structural variation, and chromothripsis in the morbid human genome. *Genome Biology*, **2017**, 18, 36	18.3	109
110	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, 210-20	11	108
109	Mutations in DCHS1 cause mitral valve prolapse. *Nature*, **2015**, 525, 109-13	50.4	107
108	Dissecting the Causal Mechanism of X-Linked Dystonia-Parkinsonism by Integrating Genome and Transcriptome Assembly. *Cell*, **2018**, 172, 897-909.e21	56.2	106
107	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, 2752-68	5.6	104
106	Haplotype-resolved diverse human genomes and integrated analysis of structural variation. *Science* , **2021**, 372,	33.3	100
105	Loss of catenin function in severe autism. *Nature*, **2015**, 520, 51-6	50.4	97
104	Cognitive influences in postural control of patients with unilateral vestibular loss. *Gait and Posture*, **2004**, 19, 105-14	2.6	89
103	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. *Nature Genetics*, **2017**, 49, 238-248	36.3	88
102	Evaluation of a susceptibility gene for schizophrenia: genotype based meta-analysis of RGS4 polymorphisms from thirteen independent samples. *Biological Psychiatry*, **2006**, 60, 152-62	7.9	80
101	Transcriptional consequences of 16p11.2 deletion and duplication in mouse cortex and multiplex autism families. *American Journal of Human Genetics*, **2014**, 94, 870-83	11	78

100	Contribution of Copy Number Variation in Idiopathic Hypogonadotropic Hypogonadism. *Journal of the Endocrine Society*, **2021**, 5, A756-A756	0.4	78
99	20.1 DISSECTING THE FUNCTIONAL CONSEQUENCES OF RECIPROCAL GENOMIC DISORDERS. *Schizophrenia Bulletin*, **2018**, 44, S33-S33	1.3	78
98	Haploinsufficiency of SOX5 at 12p12.1 is associated with developmental delays with prominent language delay, behavior problems, and mild dysmorphic features. *Human Mutation*, **2012**, 33, 728-40	4.7	62
97	Disruption of a large intergenic noncoding RNA in subjects with neurodevelopmental disabilities. *American Journal of Human Genetics*, **2012**, 91, 1128-34	11	53
96	Novel, replicated associations between dopamine D3 receptor gene polymorphisms and schizophrenia in two independent samples. *Biological Psychiatry*, **2006**, 60, 570-7	7.9	53
95	Ectopic expression of RAD52 and dn53BP1 improves homology-directed repair during CRISPR-Cas9 genome editing. *Nature Biomedical Engineering*, **2017**, 1, 878-888	19	48
94	Haploinsufficiency of KDM6A is associated with severe psychomotor retardation, global growth restriction, seizures and cleft palate. *Human Genetics*, **2013**, 132, 537-52	6.3	48
93	Autism spectrum disorder genetics: diverse genes with diverse clinical outcomes. *Harvard Review of Psychiatry*, **2014**, 22, 65-75	4.1	48
92	Whole-Genome and RNA Sequencing Reveal Variation and Transcriptomic Coordination in the Developing Human Prefrontal Cortex. *Cell Reports*, **2020**, 31, 107489	10.6	43
91	Structural Chromosomal Rearrangements Require Nucleotide-Level Resolution: Lessons from Next-Generation Sequencing in Prenatal Diagnosis. *American Journal of Human Genetics*, **2016**, 99, 1015-1033	11	43
90	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	11	42
89	The cell adhesion gene PVRL3 is associated with congenital ocular defects. *Human Genetics*, **2012**, 131, 235-50	6.3	41
88	Describing sequencing results of structural chromosome rearrangements with a suggested next-generation cytogenetic nomenclature. *American Journal of Human Genetics*, **2014**, 94, 695-709	11	40
87	Primary cilia defects causing mitral valve prolapse. *Science Translational Medicine*, **2019**, 11,	17.5	39
86	Engineering microdeletions and microduplications by targeting segmental duplications with CRISPR. *Nature Neuroscience*, **2016**, 19, 517-22	25.5	39
85	SYCP2 Translocation-Mediated Dysregulation and Frameshift Variants Cause Human Male Infertility. *American Journal of Human Genetics*, **2020**, 106, 41-57	11	39
84	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, 375-86	11	38
83	Computational Prediction of Position Effects of Apparently Balanced Human Chromosomal Rearrangements. *American Journal of Human Genetics*, **2017**, 101, 206-217	11	38

82	Htt CAG repeat expansion confers pleiotropic gains of mutant huntingtin function in chromatin regulation. *Human Molecular Genetics*, **2015**, 24, 2442-57	5.6	38
81	Paired-Duplication Signatures Mark Cryptic Inversions and Other Complex Structural Variation. *American Journal of Human Genetics*, **2015**, 97, 170-6	11	37
80	Cryptic and complex chromosomal aberrations in early-onset neuropsychiatric disorders. *American Journal of Human Genetics*, **2014**, 95, 454-61	11	37
79	Serotonin gene polymorphisms and bipolar I disorder: focus on the serotonin transporter. *Annals of Medicine*, **2005**, 37, 590-602	1.5	37
78	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. *American Journal of Human Genetics*, **2015**, 96, 784-96	11	35
77	Dopamine genes and schizophrenia: case closed or evidence pending?. *Schizophrenia Bulletin*, **2007**, 33, 1071-81	1.3	33
76	An open resource of structural variation for medical and population genetics		33
75	High coverage whole genome sequencing of the expanded 1000 Genomes Project cohort including 602 trios		31
74	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	5.3	29
73	Linkage disequilibrium patterns and functional analysis of RGS4 polymorphisms in relation to schizophrenia. *Schizophrenia Bulletin*, **2008**, 34, 118-26	1.3	29
72	Multi-platform discovery of haplotype-resolved structural variation in human genomes		26
71	Indexcov: fast coverage quality control for whole-genome sequencing. *GigaScience*, **2017**, 6, 1-6	7.6	24
70	Consanguinity associated with increased risk for bipolar I disorder in Egypt. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2009**, 150B, 879-85	3.5	23
69	Actin capping protein CAPZB regulates cell morphology, differentiation, and neural crest migration in craniofacial morphogenesis *Human Molecular Genetics*, **2016**, 25, 1255-70	5.6	22
68	Implication of LRRC4C and DPP6 in neurodevelopmental disorders. *American Journal of Medical Genetics, Part A*, **2017**, 173, 395-406	2.5	22
67	An eMERGE Clinical Center at Partners Personalized Medicine. *Journal of Personalized Medicine*, **2016**, 6,	3.6	22
66	A comprehensive genetic association and functional study of TNF in schizophrenia risk. *Schizophrenia Research*, **2006**, 83, 7-13	3.6	21
65	Large-scale exome sequencing study implicates both developmental and functional changes in the neurobiology of autism		21

64	Mutated Huntingtin Causes Testicular Pathology in Transgenic Minipig Boars. *Neurodegenerative Diseases*, **2016**, 16, 245-59	2.3	19
63	Genetic associations between neuregulin-1 SNPs and neurocognitive function in multigenerational, multiplex schizophrenia families. *Psychiatric Genetics*, **2012**, 22, 70-81	2.9	19
62	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	11	19
61	Functional annotation of rare structural variation in the human brain. *Nature Communications*, **2020**, 11, 2990	17.4	18
60	Genomic and functional overlap between somatic and germline chromosomal rearrangements. *Cell Reports*, **2014**, 9, 2001-10	10.6	18
59	Mechanisms for Structural Variation in the Human Genome. *Current Genetic Medicine Reports*, **2013**, 1, 81-90	2.2	18
58	Can RGS4 polymorphisms be viewed as credible risk factors for schizophrenia? A critical review of the evidence. *Schizophrenia Bulletin*, **2006**, 32, 203-8	1.3	18
57	Highly penetrant alterations of a critical region including BDNF in human psychopathology and obesity. *Archives of General Psychiatry*, **2012**, 69, 1238-46		16
56	Fine-mapping reveals novel alternative splicing of the dopamine transporter. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2010**, 153B, 1434-47	3.5	16
55	RGS4 polymorphisms associated with variability of cognitive performance in a family-based schizophrenia sample. *Schizophrenia Bulletin*, **2010**, 36, 983-90	1.3	15
54	Systematic association studies of mitochondrial DNA variations in schizophrenia: focus on the ND5 gene. *Schizophrenia Bulletin*, **2008**, 34, 458-65	1.3	15
53	Loss of MAGEL2 in Prader-Willi syndrome leads to decreased secretory granule and neuropeptide production. *JCI Insight*, **2020**, 5,	9.9	15
52	WNT/β-Catenin Pathway and Epigenetic Mechanisms Regulate the Pitt-Hopkins Syndrome and Schizophrenia Risk Gene. *Molecular Neuropsychiatry*, **2017**, 3, 53-71	4.9	14
51	TSC patient-derived isogenic neural progenitor cells reveal altered early neurodevelopmental phenotypes and rapamycin-induced MNK-eIF4E signaling. *Molecular Autism*, **2020**, 11, 2	6.5	14
50	Kctd13-deficient mice display short-term memory impairment and sex-dependent genetic interactions. *Human Molecular Genetics*, **2019**, 28, 1474-1486	5.6	14
49	New gene discoveries highlight functional convergence in autism and related neurodevelopmental disorders. *Current Opinion in Genetics and Development*, **2020**, 65, 195-206	4.9	13
48	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	11	13
47	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	11	13

46	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, 560-9	3.5	12
45	Evidence for secondary-variant genetic burden and non-random distribution across biological modules in a recessive ciliopathy. *Nature Genetics*, **2020**, 52, 1145-1150	36.3	12
44	Estrogen-related receptor gamma implicated in a phenotype including hearing loss and mild developmental delay. *European Journal of Human Genetics*, **2016**, 24, 1622-1626	5.3	12
43	Prioritization of genes driving congenital phenotypes of patients with de novo genomic structural variants. *Genome Medicine*, **2019**, 11, 79	14.4	11
42	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,	5.6	11
41	Mosaic copy number variation in schizophrenia. *European Journal of Human Genetics*, **2013**, 21, 1007-11	5.3	10
40	Introduction of genomics into prenatal diagnostics. *Lancet, The*, **2019**, 393, 719-721	40	9
39	Lack of association of rare functional variants in TSC1/TSC2 genes with autism spectrum disorder. *Molecular Autism*, **2013**, 4, 5	6.5	9
38	Functional analysis of upstream common polymorphisms of the dopamine transporter gene. *Schizophrenia Bulletin*, **2010**, 36, 977-82	1.3	9
37	CNView: a visualization and annotation tool for copy number variation from whole-genome sequencing		9
36	Whole exome sequencing analyses reveal gene-microbiota interactions in the context of IBD. *Gut*, **2021**, 70, 285-296	19.2	9
35	Potential molecular consequences of transgene integration: The R6/2 mouse example. *Scientific Reports*, **2017**, 7, 41120	4.9	8
34	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	5.3	8
33	Biallelic mutation of FBXL7 suggests a novel form of Hennekam syndrome. *American Journal of Medical Genetics, Part A*, **2020**, 182, 189-194	2.5	7
32	Complex and Dynamic Chromosomal Rearrangements in a Family With Seemingly Non-Mendelian Inheritance of Dopa-Responsive Dystonia. *JAMA Neurology*, **2017**, 74, 806-812	17.2	6
31	Hypomorphic mutation of the mouse Huntington's disease gene orthologue. *PLoS Genetics*, **2019**, 15, e1007765	6	6
30	Histone deacetylase knockouts modify transcription, CAG instability and nuclear pathology in Huntington disease mice. *ELife*, **2020**, 9,	8.9	6
29	Limited contribution of rare, noncoding variation to autism spectrum disorder from sequencing of 2,076 genomes in quartet families		6

28	Mapping and phasing of structural variation in patient genomes using nanopore sequencing		6
27	A novel microduplication of ARID1B: Clinical, genetic, and proteomic findings. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2478-2484	2.5	5
26	Centers for Mendelian Genomics: A decade of facilitating gene discovery.. *Genetics in Medicine*, **2022**,	8.1	5
25	Familial thrombocytopenia due to a complex structural variant resulting in a WAC-ANKRD26 fusion transcript. *Journal of Experimental Medicine*, **2021**, 218,	16.6	5
24	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	17.4	5
23	Genome-encoded cytoplasmic double-stranded RNAs, found in ALS-FTD brain, propagate neuronal loss. *Science Translational Medicine*, **2021**, 13,	17.5	5
22	A cross-disorder dosage sensitivity map of the human genome		5
21	Rare coding variation illuminates the allelic architecture, risk genes, cellular expression patterns, and phenotypic context of autism		4
20	Prevalence and phenotypic impact of rare potentially damaging variants in autism spectrum disorder. *Molecular Autism*, **2021**, 12, 65	6.5	4
19	Genome-Encoded Cytoplasmic Double-Stranded RNAs, Found in C9ORF72 ALS-FTD Brain, Provoke Propagated Neuronal Death		4
18	Transcriptional consequences of MBD5 disruption in mouse brain and CRISPR-derived neurons. *Molecular Autism*, **2020**, 11, 45	6.5	3
17	Expectations and blind spots for structural variation detection from short-read alignment and long-read assembly		3
16	De novo assembly of 64 haplotype-resolved human genomes of diverse ancestry and integrated analysis of structural variation		3
15	SYCP2 translocation-mediated dysregulation and frameshift variants cause human male infertility		3
14	Functional annotation of rare structural variation in the human brain		3
13	The female protective effect against autism spectrum disorder		3
12	A deep learning approach to identify gene targets of a therapeutic for human splicing disorders. *Nature Communications*, **2021**, 12, 3332	17.4	3
11	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. *Nature*, **2021**, 597, E3-E4	50.4	3

10	Role of the Chromosome Architectural Factor SMCHD1 in X-Chromosome Inactivation, Gene Regulation, and Disease in Humans. *Genetics*, **2019**, 213, 685-703	4	2
9	Systematic evaluation of genome sequencing as a first-tier diagnostic test for prenatal and pediatric disorders		2
8	Parallelized engineering of mutational models using piggyBac transposon delivery of CRISPR libraries		2
7	Age dependent association of inbreeding with risk for schizophrenia in Egypt. *Schizophrenia Research*, **2020**, 216, 450-459	3.6	1
6	The Role of Attention in Vestibular Processing. *Proceedings of the Human Factors and Ergonomics Society*, **2002**, 46, 255-259	0.4	1
5	Dystonia-specific mutations in THAP1 alter transcription of genes associated with neurodevelopment and myelin. *American Journal of Human Genetics*, **2021**, 108, 2145-2158	11	1
4	Prioritization of genes driving congenital phenotypes of patients with de novo genomic structural variants		1
3	De novo structural mutation rates and gamete-of-origin biases revealed through genome sequencing of 2,396 families		1
2	Xenopus models suggest convergence of gene signatures on neurogenesis in autism. *Neuron*, **2021**, 109, 743-745	13.9	0
1	Next Generation Sequencing of Prenatal Structural Chromosomal Rearrangements Using Large-Insert Libraries. *Methods in Molecular Biology*, **2019**, 1885, 251-265	1.4	