Michael E Talkowski

List of Publications by Year in Descending Order

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	Centers for Mendelian Genomics: A decade of facilitating gene discovery.. *Genetics in Medicine*, **2022**,	8.1	5
134	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
133	Prevalence and phenotypic impact of rare potentially damaging variants in autism spectrum disorder. *Molecular Autism*, **2021**, 12, 65	6.5	4
132	Whole exome sequencing analyses reveal gene-microbiota interactions in the context of IBD. *Gut*, **2021**, 70, 285-296	19.2	9
131	Xenopus models suggest convergence of gene signatures on neurogenesis in autism. *Neuron*, **2021**, 109, 743-745	13.9	0
130	Familial thrombocytopenia due to a complex structural variant resulting in a WAC-ANKRD26 fusion transcript. *Journal of Experimental Medicine*, **2021**, 218,	16.6	5
129	Haplotype-resolved diverse human genomes and integrated analysis of structural variation. *Science* , **2021**, 372,	33.3	100
128	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
127	Contribution of Copy Number Variation in Idiopathic Hypogonadotropic Hypogonadism. *Journal of the Endocrine Society*, **2021**, 5, A756-A756	0.4	78
126	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
125	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
124	A deep learning approach to identify gene targets of a therapeutic for human splicing disorders. *Nature Communications*, **2021**, 12, 3332	17.4	3
123	Genome-encoded cytoplasmic double-stranded RNAs, found in ALS-FTD brain, propagate neuronal loss. *Science Translational Medicine*, **2021**, 13,	17.5	5
122	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. *Nature*, **2021**, 597, E3-E4	50.4	3
121	The mutational constraint spectrum quantified from variation in 141,456 humans. *Nature*, **2020**, 581, 434-443	50.4	2278
120	A structural variation reference for medical and population genetics. *Nature*, **2020**, 581, 444-451	50.4	223
119	Functional annotation of rare structural variation in the human brain. *Nature Communications*, **2020**, 11, 2990	17.4	18

118	Transcriptional consequences of MBD5 disruption in mouse brain and CRISPR-derived neurons. *Molecular Autism*, **2020**, 11, 45	6.5	3
117	Age dependent association of inbreeding with risk for schizophrenia in Egypt. *Schizophrenia Research*, **2020**, 216, 450-459	3.6	1
116	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
115	Whole-Genome and RNA Sequencing Reveal Variation and Transcriptomic Coordination in the Developing Human Prefrontal Cortex. *Cell Reports*, **2020**, 31, 107489	10.6	43
114	Loss of MAGEL2 in Prader-Willi syndrome leads to decreased secretory granule and neuropeptide production. *JCI Insight*, **2020**, 5,	9.9	15
113	Histone deacetylase knockouts modify transcription, CAG instability and nuclear pathology in Huntington disease mice. *ELife*, **2020**, 9,	8.9	6
112	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
111	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
110	SYCP2 Translocation-Mediated Dysregulation and Frameshift Variants Cause Human Male Infertility. *American Journal of Human Genetics*, **2020**, 106, 41-57	11	39
109	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
108	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
107	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
106	Role of the Chromosome Architectural Factor SMCHD1 in X-Chromosome Inactivation, Gene Regulation, and Disease in Humans. *Genetics*, **2019**, 213, 685-703	4	2
105	Introduction of genomics into prenatal diagnostics. *Lancet, The*, **2019**, 393, 719-721	40	9
104	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
103	Primary cilia defects causing mitral valve prolapse. *Science Translational Medicine*, **2019**, 11,	17.5	39
102	Multi-platform discovery of haplotype-resolved structural variation in human genomes. *Nature Communications*, **2019**, 10, 1784	17.4	346
101	Hypomorphic mutation of the mouse Huntington's disease gene orthologue. *PLoS Genetics*, **2019**, 15, e1007765	6	6

100	Prioritization of genes driving congenital phenotypes of patients with de novo genomic structural variants. *Genome Medicine*, **2019**, 11, 79	14.4	11
99	Next Generation Sequencing of Prenatal Structural Chromosomal Rearrangements Using Large-Insert Libraries. *Methods in Molecular Biology*, **2019**, 1885, 251-265	1.4	
98	Kctd13-deficient mice display short-term memory impairment and sex-dependent genetic interactions. *Human Molecular Genetics*, **2019**, 28, 1474-1486	5.6	14
97	Dissecting the Causal Mechanism of X-Linked Dystonia-Parkinsonism by Integrating Genome and Transcriptome Assembly. *Cell*, **2018**, 172, 897-909.e21	56.2	106
96	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
95	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
94	Insights into clonal haematopoiesis from 8,342 mosaic chromosomal alterations. *Nature*, **2018**, 559, 350-355	36.4	144
93	20.1 DISSECTING THE FUNCTIONAL CONSEQUENCES OF RECIPROCAL GENOMIC DISORDERS. *Schizophrenia Bulletin*, **2018**, 44, S33-S33	1.3	78
92	Genome-wide de novo risk score implicates promoter variation in autism spectrum disorder. *Science* , **2018**, 362,	33.3	134
91	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
90	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
89	Potential molecular consequences of transgene integration: The R6/2 mouse example. *Scientific Reports*, **2017**, 7, 41120	4.9	8
88	Defining the diverse spectrum of inversions, complex structural variation, and chromothripsis in the morbid human genome. *Genome Biology*, **2017**, 18, 36	18.3	109
87	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
86	Ectopic expression of RAD52 and dn53BP1 improves homology-directed repair during CRISPR-Cas9 genome editing. *Nature Biomedical Engineering*, **2017**, 1, 878-888	19	48
85	Indexcov: fast coverage quality control for whole-genome sequencing. *GigaScience*, **2017**, 6, 1-6	7.6	24
84	Computational Prediction of Position Effects of Apparently Balanced Human Chromosomal Rearrangements. *American Journal of Human Genetics*, **2017**, 101, 206-217	11	38
83	A novel microduplication of ARID1B: Clinical, genetic, and proteomic findings. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2478-2484	2.5	5

82	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
81	Mapping and phasing of structural variation in patient genomes using nanopore sequencing. *Nature Communications*, **2017**, 8, 1326	17.4	191
80	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. *Nature Genetics*, **2017**, 49, 36-45	36.3	172
79	Implication of LRRC4C and DPP6 in neurodevelopmental disorders. *American Journal of Medical Genetics, Part A*, **2017**, 173, 395-406	2.5	22
78	Mutated Huntingtin Causes Testicular Pathology in Transgenic Minipig Boars. *Neurodegenerative Diseases*, **2016**, 16, 245-59	2.3	19
77	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
76	Engineering microdeletions and microduplications by targeting segmental duplications with CRISPR. *Nature Neuroscience*, **2016**, 19, 517-22	25.5	39
75	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
74	An eMERGE Clinical Center at Partners Personalized Medicine. *Journal of Personalized Medicine*, **2016**, 6,	3.6	22
73	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
72	An Ancient, Unified Mechanism for Metformin Growth Inhibition in C. elegans and Cancer. *Cell*, **2016**, 167, 1705-1718.e13	56.2	134
71	Paired-Duplication Signatures Mark Cryptic Inversions and Other Complex Structural Variation. *American Journal of Human Genetics*, **2015**, 97, 170-6	11	37
70	Loss of β-catenin function in severe autism. *Nature*, **2015**, 520, 51-6	50.4	97
69	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
68	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. *Neuron*, **2015**, 87, 1215-1233	13.9	806
67	Mutations in DCHS1 cause mitral valve prolapse. *Nature*, **2015**, 525, 109-13	50.4	107
66	Htt CAG repeat expansion confers pleiotropic gains of mutant huntingtin function in chromatin regulation. *Human Molecular Genetics*, **2015**, 24, 2442-57	5.6	38
65	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

64	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
63	Efficient ablation of genes in human hematopoietic stem and effector cells using CRISPR/Cas9. *Cell Stem Cell*, **2014**, 15, 643-52	18	324
62	Cryptic and complex chromosomal aberrations in early-onset neuropsychiatric disorders. *American Journal of Human Genetics*, **2014**, 95, 454-61	11	37
61	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
60	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
59	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
58	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
57	Genomic and functional overlap between somatic and germline chromosomal rearrangements. *Cell Reports*, **2014**, 9, 2001-10	10.6	18
56	Autism spectrum disorder genetics: diverse genes with diverse clinical outcomes. *Harvard Review of Psychiatry*, **2014**, 22, 65-75	4.1	48
55	Lack of association of rare functional variants in TSC1/TSC2 genes with autism spectrum disorder. *Molecular Autism*, **2013**, 4, 5	6.5	9
54	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
53	Mosaic copy number variation in schizophrenia. *European Journal of Human Genetics*, **2013**, 21, 1007-11	5.3	10
52	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
51	Mechanisms for Structural Variation in the Human Genome. *Current Genetic Medicine Reports*, **2013**, 1, 81-90	2.2	18
50	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
49	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
48	The cell adhesion gene PVRL3 is associated with congenital ocular defects. *Human Genetics*, **2012**, 131, 235-50	6.3	41
47	Highly penetrant alterations of a critical region including BDNF in human psychopathology and obesity. *Archives of General Psychiatry*, **2012**, 69, 1238-46		16

46	Clinical diagnosis by whole-genome sequencing of a prenatal sample. *New England Journal of Medicine*, **2012**, 367, 2226-32	59.2	144
45	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
44	KCTD13 is a major driver of mirrored neuroanatomical phenotypes of the 16p11.2 copy number variant. *Nature*, **2012**, 485, 363-7	50.4	281
43	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
42	Sequencing chromosomal abnormalities reveals neurodevelopmental loci that confer risk across diagnostic boundaries. *Cell*, **2012**, 149, 525-37	56.2	441
41	Disruption of a large intergenic noncoding RNA in subjects with neurodevelopmental disabilities. *American Journal of Human Genetics*, **2012**, 91, 1128-34	11	53
40	Genetic associations between neuregulin-1 SNPs and neurocognitive function in multigenerational, multiplex schizophrenia families. *Psychiatric Genetics*, **2012**, 22, 70-81	2.9	19
39	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
38	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
37	RGS4 polymorphisms associated with variability of cognitive performance in a family-based schizophrenia sample. *Schizophrenia Bulletin*, **2010**, 36, 983-90	1.3	15
36	Functional analysis of upstream common polymorphisms of the dopamine transporter gene. *Schizophrenia Bulletin*, **2010**, 36, 977-82	1.3	9
35	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
34	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
33	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
32	Association study of 21 circadian genes with bipolar I disorder, schizoaffective disorder, and schizophrenia. *Bipolar Disorders*, **2009**, 11, 701-10	3.8	120
31	Systematic association studies of mitochondrial DNA variations in schizophrenia: focus on the ND5 gene. *Schizophrenia Bulletin*, **2008**, 34, 458-65	1.3	15
30	A network of dopaminergic gene variations implicated as risk factors for schizophrenia. *Human Molecular Genetics*, **2008**, 17, 747-58	5.6	120
29	Linkage disequilibrium patterns and functional analysis of RGS4 polymorphisms in relation to schizophrenia. *Schizophrenia Bulletin*, **2008**, 34, 118-26	1.3	29

28	Dopamine genes and schizophrenia: case closed or evidence pending?. *Schizophrenia Bulletin*, **2007**, 33, 1071-81	1.3	33
27	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
26	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
25	Novel, replicated associations between dopamine D3 receptor gene polymorphisms and schizophrenia in two independent samples. *Biological Psychiatry*, **2006**, 60, 570-7	7.9	53
24	A comprehensive genetic association and functional study of TNF in schizophrenia risk. *Schizophrenia Research*, **2006**, 83, 7-13	3.6	21
23	Serotonin gene polymorphisms and bipolar I disorder: focus on the serotonin transporter. *Annals of Medicine*, **2005**, 37, 590-602	1.5	37
22	Cognitive influences in postural control of patients with unilateral vestibular loss. *Gait and Posture*, **2004**, 19, 105-14	2.6	89
21	The Role of Attention in Vestibular Processing. *Proceedings of the Human Factors and Ergonomics Society*, **2002**, 46, 255-259	0.4	1
20	Rare coding variation illuminates the allelic architecture, risk genes, cellular expression patterns, and phenotypic context of autism		4
19	Prioritization of genes driving congenital phenotypes of patients with de novo genomic structural variants		1
18	Systematic evaluation of genome sequencing as a first-tier diagnostic test for prenatal and pediatric disorders		2
17	CNView: a visualization and annotation tool for copy number variation from whole-genome sequencing		9
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11	De novo structural mutation rates and gamete-of-origin biases revealed through genome sequencing of 2,396 families		1

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