

# Michael E Talkowski

## List of Publications by Year in descending order

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Version: 2024-02-01

122  
papers

20,162  
citations

47006

47  
h-index

18130

120  
g-index

167  
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167  
docs citations

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times ranked

33479  
citing authors

| #  | ARTICLE   | IF   | CITATIONS |
|----|---|------|-----------|
| 1  | The mutational constraint spectrum quantified from variation in 141,456 humans. <i>Nature</i> , 2020, 581, 434-443.   | 27.8 | 6,140     |
| 2  | Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. <i>Cell</i> , 2020, 180, 568-584.e23.  | 28.9 | 1,422     |
| 3  | Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. <i>Neuron</i> , 2015, 87, 1215-1233.   | 8.1  | 1,219     |
| 4  | Multi-platform discovery of haplotype-resolved structural variation in human genomes. <i>Nature Communications</i> , 2019, 10, 1784.  | 12.8 | 636       |
| 5  | A structural variation reference for medical and population genetics. <i>Nature</i> , 2020, 581, 444-451.   | 27.8 | 614       |
| 6  | Sequencing Chromosomal Abnormalities Reveals Neurodevelopmental Loci that Confer Risk across Diagnostic Boundaries. <i>Cell</i> , 2012, 149, 525-537.   | 28.9 | 534       |
| 7  | Low Incidence of Off-Target Mutations in Individual CRISPR-Cas9 and TALEN Targeted Human Stem Cell Clones Detected by Whole-Genome Sequencing. <i>Cell Stem Cell</i> , 2014, 15, 27-30.   | 11.1 | 456       |
| 8  | Efficient Ablation of Genes in Human Hematopoietic Stem and Effector Cells using CRISPR/Cas9. <i>Cell Stem Cell</i> , 2014, 15, 643-652.  | 11.1 | 406       |
| 9  | KCTD13 is a major driver of mirrored neuroanatomical phenotypes of the 16p11.2 copy number variant. <i>Nature</i> , 2012, 485, 363-367.   | 27.8 | 363       |
| 10 | Haplotype-resolved diverse human genomes and integrated analysis of structural variation. <i>Science</i> , 2021, 372, .   | 12.6 | 358       |
| 11 | Mapping and phasing of structural variation in patient genomes using nanopore sequencing. <i>Nature Communications</i> , 2017, 8, 1326.   | 12.8 | 315       |
| 12 | <i>CHD8</i> regulates neurodevelopmental pathways associated with autism spectrum disorder in neural progenitors. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, E4468-77. | 7.1  | 297       |
| 13 | Insights into clonal haematopoiesis from 8,342 mosaic chromosomal alterations. <i>Nature</i> , 2018, 559, 350-355.  | 27.8 | 279       |
| 14 | The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. <i>Nature Genetics</i> , 2017, 49, 36-45.   | 21.4 | 251       |
| 15 | An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. <i>Nature Genetics</i> , 2018, 50, 727-736.  | 21.4 | 235       |
| 16 | Genome-wide de novo risk score implicates promoter variation in autism spectrum disorder. <i>Science</i> , 2018, 362, .   | 12.6 | 234       |
| 17 | Complex reorganization and predominant non-homologous repair following chromosomal breakage in karyotypically balanced germline rearrangements and transgenic integration. <i>Nature Genetics</i> , 2012, 44, 390-397.          | 21.4 | 229       |
| 18 | Whole-Genome Sequencing to Characterize Monogenic and Polygenic Contributions in Patients Hospitalized With Early-Onset Myocardial Infarction. <i>Circulation</i> , 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. <i>American Journal of Human Genetics</i> , 2011, 89, 551-563. | 6.2  | 195       |
| 20 | An Ancient, Unified Mechanism for Metformin Growth Inhibition in <i>C.Âlegans</i> and Cancer. <i>Cell</i> , 2016, 167, 1705-1718.e13.   | 28.9 | 181       |
| 21 | Clinical Diagnosis by Whole-Genome Sequencing of a Prenatal Sample. <i>New England Journal of Medicine</i> , 2012, 367, 2226-2232.  | 27.0 | 174       |
| 22 | Dissecting the Causal Mechanism of X-Linked Dystonia-Parkinsonism by Integrating Genome and Transcriptome Assembly. <i>Cell</i> , 2018, 172, 897-909.e21.   | 28.9 | 163       |
| 23 | Defining the diverse spectrum of inversions, complex structural variation, and chromothripsis in the morbid humanÂgenome. <i>Genome Biology</i> , 2017, 18, 36.   | 8.8  | 159       |
| 24 | Next-Generation Sequencing Strategies Enable Routine Detection of Balanced Chromosome Rearrangements for Clinical Diagnostics and Genetic Research. <i>American Journal of Human Genetics</i> , 2011, 88, 469-481.      | 6.2  | 154       |
| 25 | Mutations in DCHS1 cause mitral valve prolapse. <i>Nature</i> , 2015, 525, 109-113.   | 27.8 | 150       |
| 26 | Loss of Î-catenin function in severe autism. <i>Nature</i> , 2015, 520, 51-56.  | 27.8 | 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. <i>Human Molecular Genetics</i> , 2014, 23, 2752-2768.            | 2.9  | 140       |
| 28 | Exonic Deletions in AUTS2 Cause a Syndromic Form of Intellectual Disability and Suggest a Critical Role for the C Terminus. <i>American Journal of Human Genetics</i> , 2013, 92, 210-220.                              | 6.2  | 135       |
| 29 | Association study of 21 circadian genes with bipolar I disorder, schizoaffective disorder, and schizophrenia. <i>Bipolar Disorders</i> , 2009, 11, 701-710.   | 1.9  | 133       |
| 30 | SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. <i>Nature Genetics</i> , 2017, 49, 238-248.                                       | 21.4 | 131       |
| 31 | A network of dopaminergic gene variations implicated as risk factors for schizophrenia. <i>Human Molecular Genetics</i> , 2008, 17, 747-758.  | 2.9  | 124       |
| 32 | Transcriptional Consequences of 16p11.2 Deletion and Duplication in Mouse Cortex and Multiplex Autism Families. <i>American Journal of Human Genetics</i> , 2014, 94, 870-883.  | 6.2  | 116       |
| 33 | Cognitive influences in postural control of patients with unilateral vestibular loss. <i>Gait and Posture</i> , 2004, 19, 105-114.  | 1.4  | 113       |
| 34 | Whole-Genome and RNA Sequencing Reveal Variation and Transcriptomic Coordination in the Developing Human Prefrontal Cortex. <i>Cell Reports</i> , 2020, 31, 107489.   | 6.4  | 91        |
| 35 | Evaluation of a Susceptibility Gene for Schizophrenia: Genotype Based Meta-Analysis of RGS4 Polymorphisms from Thirteen Independent Samples. <i>Biological Psychiatry</i> , 2006, 60, 152-162.                          | 1.3  | 87        |
| 36 | Haploinsufficiency of <i>SOX5</i> at 12p12.1 is associated with developmental delays with prominent language delay, behavior problems, and mild dysmorphic features. <i>Human Mutation</i> , 2012, 33, 728-740.         | 2.5  | 85        |

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|----|--|------|-----------|
| 37 | Ectopic expression of RAD52 and dn53BP1 improves homology-directed repair during CRISPR-Cas9 genome editing. <i>Nature Biomedical Engineering</i> , 2017, 1, 878-888.  | 22.5 | 83        |
| 38 | Primary cilia defects causing mitral valve prolapse. <i>Science Translational Medicine</i> , 2019, 11, .   | 12.4 | 76        |
| 39 | Engineering microdeletions and microduplications by targeting segmental duplications with CRISPR. <i>Nature Neuroscience</i> , 2016, 19, 517-522.  | 14.8 | 72        |
| 40 | Expectations and blind spots for structural variation detection from long-read assemblies and short-read genome sequencing technologies. <i>American Journal of Human Genetics</i> , 2021, 108, 919-928.     | 6.2  | 72        |
| 41 | SYCP2 Translocation-Mediated Dysregulation and Frameshift Variants Cause Human Male Infertility. <i>American Journal of Human Genetics</i> , 2020, 106, 41-57.   | 6.2  | 66        |
| 42 | Novel, Replicated Associations Between Dopamine D3 Receptor Gene Polymorphisms and Schizophrenia in Two Independent Samples. <i>Biological Psychiatry</i> , 2006, 60, 570-577.                               | 1.3  | 62        |
| 43 | Disruption of a Large Intergenic Noncoding RNA in Subjects with Neurodevelopmental Disabilities. <i>American Journal of Human Genetics</i> , 2012, 91, 1128-1134.  | 6.2  | 61        |
| 44 | Haploinsufficiency of KDM6A is associated with severe psychomotor retardation, global growth restriction, seizures and cleft palate. <i>Human Genetics</i> , 2013, 132, 537-552.                             | 3.8  | 60        |
| 45 | Translocations Disrupting PHF21A in the Potocki-Shaffer-Syndrome Region Are Associated with Intellectual Disability and Craniofacial Anomalies. <i>American Journal of Human Genetics</i> , 2012, 91, 56-72. | 6.2  | 59        |
| 46 | Autism Spectrum Disorder Genetics. <i>Harvard Review of Psychiatry</i> , 2014, 22, 65-75.  | 2.1  | 59        |
| 47 | De novo structural mutation rates and gamete-of-origin biases revealed through genome sequencing of 2,396 families. <i>American Journal of Human Genetics</i> , 2021, 108, 597-607.                          | 6.2  | 57        |
| 48 | Htt CAG repeat expansion confers pleiotropic gains of mutant huntingtin function in chromatin regulation. <i>Human Molecular Genetics</i> , 2015, 24, 2442-2457.   | 2.9  | 53        |
| 49 | A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. <i>American Journal of Human Genetics</i> , 2015, 96, 784-796.  | 6.2  | 53        |
| 50 | Structural Chromosomal Rearrangements Require Nucleotide-Level Resolution: Lessons from Next-Generation Sequencing in Prenatal Diagnosis. <i>American Journal of Human Genetics</i> , 2016, 99, 1015-1033.   | 6.2  | 53        |
| 51 | Computational Prediction of Position Effects of Apparently Balanced Human Chromosomal Rearrangements. <i>American Journal of Human Genetics</i> , 2017, 101, 206-217.  | 6.2  | 51        |
| 52 | The cell adhesion gene PVRL3 is associated with congenital ocular defects. <i>Human Genetics</i> , 2012, 131, 235-250.   | 3.8  | 46        |
| 53 | Cryptic and Complex Chromosomal Aberrations in Early-Onset Neuropsychiatric Disorders. <i>American Journal of Human Genetics</i> , 2014, 95, 454-461.  | 6.2  | 45        |
| 54 | Paired-Duplication Signatures Mark Cryptic Inversions and Other Complex Structural Variation. <i>American Journal of Human Genetics</i> , 2015, 97, 170-176.   | 6.2  | 45        |

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|----|---|------|-----------|
| 55 | Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. <i>Nature</i> , 2021, 597, E3-E4.   | 27.8 | 45        |
| 56 | Centers for Mendelian Genomics: A decade of facilitating gene discovery. <i>Genetics in Medicine</i> , 2022, 24, 784-797.   | 2.4  | 44        |
| 57 | Molecular Analysis of a Deletion Hotspot in the NRXN1 Region Reveals the Involvement of Short Inverted Repeats in Deletion CNVs. <i>American Journal of Human Genetics</i> , 2013, 92, 375-386.   | 6.2  | 42        |
| 58 | Describing Sequencing Results of Structural Chromosome Rearrangements with a Suggested Next-Generation Cytogenetic Nomenclature. <i>American Journal of Human Genetics</i> , 2014, 94, 695-709.   | 6.2  | 42        |
| 59 | Reciprocal deletion and duplication at 2q23.1 indicates a role for MBD5 in autism spectrum disorder. <i>European Journal of Human Genetics</i> , 2014, 22, 57-63.                                 | 2.8  | 42        |
| 60 | Implication of <i>LRRC4C</i> and <i>DPP6</i> in neurodevelopmental disorders. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 395-406.   | 1.2  | 40        |
| 61 | Loss of MAGEL2 in Prader-Willi syndrome leads to decreased secretory granule and neuropeptide production. <i>JCI Insight</i> , 2020, 5, .   | 5.0  | 40        |
| 62 | Serotonin gene polymorphisms and bipolar I disorder: Focus on the serotonin transporter. <i>Annals of Medicine</i> , 2005, 37, 590-602.   | 3.8  | 39        |
| 63 | Dopamine Genes and Schizophrenia: Case Closed or Evidence Pending?. <i>Schizophrenia Bulletin</i> , 2007, 33, 1071-1081.  | 4.3  | 37        |
| 64 | Indexcov: fast coverage quality control for whole-genome sequencing. <i>GigaScience</i> , 2017, 6, 1-6.   | 6.4  | 36        |
| 65 | 16p11.2 deletion is associated with hyperactivation of human iPSC-derived dopaminergic neuron networks and is rescued by RHOA inhibition in vitro. <i>Nature Communications</i> , 2021, 12, 2897. | 12.8 | 35        |
| 66 | Linkage Disequilibrium Patterns and Functional Analysis of RGS4 Polymorphisms in Relation to Schizophrenia. <i>Schizophrenia Bulletin</i> , 2007, 34, 118-126.                                    | 4.3  | 34        |
| 67 | Kctd13-deficient mice display short-term memory impairment and sex-dependent genetic interactions. <i>Human Molecular Genetics</i> , 2019, 28, 1474-1486.   | 2.9  | 32        |
| 68 | Functional annotation of rare structural variation in the human brain. <i>Nature Communications</i> , 2020, 11, 2990.   | 12.8 | 32        |
| 69 | An eMERGE Clinical Center at Partners Personalized Medicine. <i>Journal of Personalized Medicine</i> , 2016, 6, 5.  | 2.5  | 31        |
| 70 | Actin capping protein CAPZB regulates cell morphology, differentiation, and neural crest migration in craniofacial morphogenesis. <i>Human Molecular Genetics</i> , 2016, 25, 1255-1270.          | 2.9  | 30        |
| 71 | The female protective effect against autism spectrum disorder. <i>Cell Genomics</i> , 2022, 2, 100134.  | 6.5  | 30        |
| 72 | Mechanisms for Structural Variation in the Human Genome. <i>Current Genetic Medicine Reports</i> , 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. <i>American Journal of Human Genetics</i> , 2018, 102, 1090-1103. | 6.2  | 29        |
| 74 | TSC patient-derived isogenic neural progenitor cells reveal altered early neurodevelopmental phenotypes and rapamycin-induced MNK-eIF4E signaling. <i>Molecular Autism</i> , 2020, 11, 2.             | 4.9  | 29        |
| 75 | Consanguinity associated with increased risk for bipolar I disorder in Egypt. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2009, 150B, 879-885.                    | 1.7  | 28        |
| 76 | A Balanced Translocation in Kallmann Syndrome Implicates a Long Noncoding RNA, RMST, as a GnRH Neuronal Regulator. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, e231-e244.    | 3.6  | 28        |
| 77 | New gene discoveries highlight functional convergence in autism and related neurodevelopmental disorders. <i>Current Opinion in Genetics and Development</i> , 2020, 65, 195-206.                     | 3.3  | 27        |
| 78 | Genome-encoded cytoplasmic double-stranded RNAs, found in <i>C9ORF72</i> ALS-FTD brain, propagate neuronal loss. <i>Science Translational Medicine</i> , 2021, 13, .                                  | 12.4 | 27        |
| 79 | Whole exome sequencing analyses reveal gene-microbiota interactions in the context of IBD. <i>Gut</i> , 2021, 70, gutjnl-2019-319706.   | 12.1 | 26        |
| 80 | A deep learning approach to identify gene targets of a therapeutic for human splicing disorders. <i>Nature Communications</i> , 2021, 12, 3332.   | 12.8 | 26        |
| 81 | Genetic associations between neuregulin-1 SNPs and neurocognitive function in multigenerational, multiplex schizophrenia families. <i>Psychiatric Genetics</i> , 2012, 22, 70-81.                     | 1.1  | 23        |
| 82 | Highly Penetrant Alterations of a Critical Region Including BDNF in Human Psychopathology and Obesity. <i>Archives of General Psychiatry</i> , 2012, 69, 1238.  | 12.3 | 22        |
| 83 | Mutated Huntingtin Causes Testicular Pathology in Transgenic Minipig Boars. <i>Neurodegenerative Diseases</i> , 2016, 16, 245-259.  | 1.4  | 22        |
| 84 | Evidence for secondary-variant genetic burden and non-random distribution across biological modules in a recessive ciliopathy. <i>Nature Genetics</i> , 2020, 52, 1145-1150.                          | 21.4 | 22        |
| 85 | Prevalence and phenotypic impact of rare potentially damaging variants in autism spectrum disorder. <i>Molecular Autism</i> , 2021, 12, 65.   | 4.9  | 22        |
| 86 | A comprehensive genetic association and functional study of TNF in schizophrenia risk. <i>Schizophrenia Research</i> , 2006, 83, 7-13.  | 2.0  | 21        |
| 87 | Genomic and Functional Overlap between Somatic and Germline Chromosomal Rearrangements. <i>Cell Reports</i> , 2014, 9, 2001-2010.   | 6.4  | 21        |
| 88 | Familial thrombocytopenia due to a complex structural variant resulting in a <i>WAC-ANKRD26</i> fusion transcript. <i>Journal of Experimental Medicine</i> , 2021, 218, .                             | 8.5  | 20        |
| 89 | Can RGS4 Polymorphisms Be Viewed as Credible Risk Factors for Schizophrenia? A Critical Review of the Evidence. <i>Schizophrenia Bulletin</i> , 2006, 32, 203-208.                                    | 4.3  | 19        |
| 90 | WNT/ $\beta$ -Catenin Pathway and Epigenetic Mechanisms Regulate the Pitt-Hopkins Syndrome and Schizophrenia Risk Gene TCF4. <i>Molecular Neuropsychiatry</i> , 2017, 3, 53-71.                       | 2.9  | 19        |

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|-----|---|------|-----------|
| 91  | Prioritization of genes driving congenital phenotypes of patients with de novo genomic structural variants. <i>Genome Medicine</i> , 2019, 11, 79.  | 8.2  | 19        |
| 92  | Systematic Association Studies of Mitochondrial DNA Variations in Schizophrenia: Focus on the ND5 Gene. <i>Schizophrenia Bulletin</i> , 2008, 34, 458-465.  | 4.3  | 18        |
| 93  | Fine-mapping reveals novel alternative splicing of the dopamine transporter. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2010, 153B, 1434-1447.   | 1.7  | 18        |
| 94  | RGS4 Polymorphisms Associated With Variability of Cognitive Performance in a Family-Based Schizophrenia Sample. <i>Schizophrenia Bulletin</i> , 2010, 36, 983-990.  | 4.3  | 18        |
| 95  | Lack of association of rare functional variants in TSC1/TSC2 genes with autism spectrum disorder. <i>Molecular Autism</i> , 2013, 4, 5.   | 4.9  | 16        |
| 96  | Convergent patterns of association between phenylalanine hydroxylase variants and schizophrenia in four independent samples. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2009, 150B, 560-569. | 1.7  | 15        |
| 97  | Mosaic copy number variation in schizophrenia. <i>European Journal of Human Genetics</i> , 2013, 21, 1007-1011.   | 2.8  | 15        |
| 98  | Potential molecular consequences of transgene integration: The R6/2 mouse example. <i>Scientific Reports</i> , 2017, 7, 41120.  | 3.3  | 14        |
| 99  | Introduction of genomics into prenatal diagnostics. <i>Lancet, The</i> , 2019, 393, 719-721.  | 13.7 | 13        |
| 100 | Hypomorphic mutation of the mouse Huntington's disease gene orthologue. <i>PLoS Genetics</i> , 2019, 15, e1007765.  | 3.5  | 13        |
| 101 | Biallelic mutation of FBXL7 suggests a novel form of Hennekam syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 189-194.   | 1.2  | 13        |
| 102 | Dystonia-specific mutations in THAP1 alter transcription of genes associated with neurodevelopment and myelin. <i>American Journal of Human Genetics</i> , 2021, 108, 2145-2158.  | 6.2  | 13        |
| 103 | Estrogen-related receptor gamma implicated in a phenotype including hearing loss and mild developmental delay. <i>European Journal of Human Genetics</i> , 2016, 24, 1622-1626.   | 2.8  | 12        |
| 104 | Transcriptional consequences of MBD5 disruption in mouse brain and CRISPR-derived neurons. <i>Molecular Autism</i> , 2020, 11, 45.  | 4.9  | 11        |
| 105 | Orgo-Seq integrates single-cell and bulk transcriptomic data to identify cell type specific-driver genes associated with autism spectrum disorder. <i>Nature Communications</i> , 2022, 13, .                                     | 12.8 | 11        |
| 106 | Functional Analysis of Upstream Common Polymorphisms of the Dopamine Transporter Gene. <i>Schizophrenia Bulletin</i> , 2010, 36, 977-982.   | 4.3  | 10        |
| 107 | Prevalence and Phenotypic Effects of Copy Number Variants in Isolated Hypogonadotropic Hypogonadism. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, 2228-2242.  | 3.6  | 10        |
| 108 | Histone deacetylase knockouts modify transcription, CAG instability and nuclear pathology in Huntington disease mice. <i>ELife</i> , 2020, 9, .   | 6.0  | 9         |

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|-----|--|-----|-----------|
| 109 | Phenotypic interpretation of complex chromosomal rearrangements informed by nucleotide-level resolution and structural organization of chromatin. <i>European Journal of Human Genetics</i> , 2018, 26, 374-381. | 2.8 | 8         |
| 110 | A neurodevelopmental disorder caused by a novel de novo SVA insertion in exon 13 of the SRCAP gene. <i>European Journal of Human Genetics</i> , 2022, 30, 1083-1087.   | 2.8 | 8         |
| 111 | Complex and Dynamic Chromosomal Rearrangements in a Family With Seemingly Non-Mendelian Inheritance of Dopa-Responsive Dystonia. <i>JAMA Neurology</i> , 2017, 74, 806.  | 9.0 | 7         |
| 112 | A novel microduplication of <i>ARID1B</i> : Clinical, genetic, and proteomic findings. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2478-2484.   | 1.2 | 7         |
| 113 | Developmental regulation of neuronal gene expression by Elongator complex protein 1 dosage. <i>Journal of Genetics and Genomics</i> , 2022, 49, 654-665.   | 3.9 | 6         |
| 114 | Role of the Chromosome Architectural Factor SMCHD1 in X-Chromosome Inactivation, Gene Regulation, and Disease in Humans. <i>Genetics</i> , 2019, 213, 685-703.   | 2.9 | 5         |
| 115 | The Role of Attention in Vestibular Processing. <i>Proceedings of the Human Factors and Ergonomics Society</i> , 2002, 46, 255-259.  | 0.3 | 1         |
| 116 | Age dependent association of inbreeding with risk for schizophrenia in Egypt. <i>Schizophrenia Research</i> , 2020, 216, 450-459.  | 2.0 | 1         |
| 117 | <i>Xenopus</i> models suggest convergence of gene signatures on neurogenesis in autism. <i>Neuron</i> , 2021, 109, 743-745.  | 8.1 | 1         |
| 118 | Cover Image, Volume 173A, Number 2, February 2017. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, i.   | 1.2 | 0         |
| 119 | 20.1 DISSECTING THE FUNCTIONAL CONSEQUENCES OF RECIPROCAL GENOMIC DISORDERS. <i>Schizophrenia Bulletin</i> , 2018, 44, S33-S33.  | 4.3 | 0         |
| 120 | Next Generation Sequencing of Prenatal Structural Chromosomal Rearrangements Using Large-Insert Libraries. <i>Methods in Molecular Biology</i> , 2019, 1885, 251-265.  | 0.9 | 0         |
| 121 | Contribution of Copy Number Variation in Idiopathic Hypogonadotropic Hypogonadism. <i>Journal of the Endocrine Society</i> , 2021, 5, A756-A756.   | 0.2 | 0         |
| 122 | Dissecting the Causal Mechanism of X-Linked Dystonia-Parkinsonism by Integrating Genome and Transcriptome Assembly. <i>SSRN Electronic Journal</i> , 0, , .  | 0.4 | 0         |