Timothy W Yu

List of Publications by Year
in descending order

82 papers	18,794 citations	41323 49 h-index	60583 81 g-index
91 all docs	91 docs citations	91 times ranked	25835 citing authors

#	Article	IF	Citations
1	Synaptic, transcriptional and chromatin genes disrupted in autism. Nature, 2014, 515, 209-215.	13.7	2,254
2	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. Nature Genetics, 2013, 45, 984-994.	9.4	2,067
3	Dentate Granule Cell Neurogenesis Is Increased by Seizures and Contributes to Aberrant Network Reorganization in the Adult Rat Hippocampus. Journal of Neuroscience, 1997, 17, 3727-3738.	1.7	1,744
4	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
5	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. Neuron, 2015, 87, 1215-1233.	3.8	1,219
6	Multiple Recurrent De Novo CNVs, Including Duplications of the 7q11.23 Williams Syndrome Region, Are Strongly Associated with Autism. Neuron, 2011, 70, 863-885.	3.8	1,146
7	Psychiatric genome-wide association study analyses implicate neuronal, immune and histone pathways. Nature Neuroscience, 2015, 18, 199-209.	7.1	701
8	Meta-analysis of GWAS of over 16,000 individuals with autism spectrum disorder highlights a novel locus at 10q24.32 and a significant overlap with schizophrenia. Molecular Autism, 2017, 8, 21.	2.6	495
9	Patient-Customized Oligonucleotide Therapy for a Rare Genetic Disease. New England Journal of Medicine, 2019, 381, 1644-1652.	13.9	481
10	Polygenic transmission disequilibrium confirms that common and rare variation act additively to create risk for autism spectrum disorders. Nature Genetics, 2017, 49, 978-985.	9.4	401
11	Using Whole-Exome Sequencing to Identify Inherited Causes of Autism. Neuron, 2013, 77, 259-273.	3.8	383
12	A proprotein convertase subtilisin/kexin type 9 neutralizing antibody reduces serum cholesterol in mice and nonhuman primates. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 9820-9825.	3.3	372
13	Common genetic variants, acting additively, are a major source of risk for autism. Molecular Autism, 2012, 3, 9.	2.6	357
14	Somatic Mutations in Cerebral Cortical Malformations. New England Journal of Medicine, 2014, 371, 733-743.	13.9	326
15	Dynamic regulation of axon guidance. Nature Neuroscience, 2001, 4, 1169-1176.	7.1	294
16	Mutations in WDR62, encoding a centrosome-associated protein, cause microcephaly with simplified gyri and abnormal cortical architecture. Nature Genetics, 2010, 42, 1015-1020.	9.4	259
17	Exome and genome sequencing for pediatric patients with congenital anomalies or intellectual disability: an evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 2029-2037.	1.1	229
18	Joint Analysis of Psychiatric Disorders Increases Accuracy of Risk Prediction for Schizophrenia, Bipolar Disorder, and Major Depressive Disorder. American Journal of Human Genetics, 2015, 96, 283-294.	2.6	225

#	Article	IF	Citations
19	The Netrin Receptor UNC-40/DCC Stimulates Axon Attraction and Outgrowth through Enabled and, in Parallel, Rac and UNC-115/AbLIM. Neuron, 2003, 37, 53-65.	3.8	216
20	C. elegans Slit Acts in Midline, Dorsal-Ventral, and Anterior-Posterior Guidance via the SAX-3/Robo Receptor. Neuron, 2001, 32, 25-38.	3.8	209
21	The ion channel TRPV1 regulates the activation and proinflammatory properties of CD4+ T cells. Nature Immunology, 2014, 15, 1055-1063.	7.0	193
22	Interpretation of Genomic Sequencing Results in Healthy and Ill Newborns: Results from the BabySeq Project. American Journal of Human Genetics, 2019, 104, 76-93.	2.6	176
23	Newborn Sequencing in Genomic Medicine and Public Health. Pediatrics, 2017, 139, .	1.0	174
24	Exome Sequencing and Functional Validation in Zebrafish Identify GTDC2 Mutations as a Cause of Walker-Warburg Syndrome. American Journal of Human Genetics, 2012, 91, 541-547.	2.6	167
25	Whole-Exome Sequencing and Homozygosity Analysis Implicate Depolarization-Regulated Neuronal Genes in Autism. PLoS Genetics, 2012, 8, e1002635.	1.5	164
26	Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. Npj Genomic Medicine, 2019, 4, 19.	1.7	163
27	Identification of KLHL41 Mutations Implicates BTB-Kelch-Mediated Ubiquitination as an Alternate Pathway to Myofibrillar Disruption in Nemaline Myopathy. American Journal of Human Genetics, 2013, 93, 1108-1117.	2.6	147
28	Shared receptors in axon guidance: SAX-3/Robo signals via UNC-34/Enabled and a Netrin-independent UNC-40/DCC function. Nature Neuroscience, 2002, 5, 1147-1154.	7.1	144
29	A Genome-wide Association Study of Autism Using the Simons Simplex Collection: Does Reducing Phenotypic Heterogeneity in Autism Increase Genetic Homogeneity?. Biological Psychiatry, 2015, 77, 775-784.	0.7	133
30	Thrombotic Intracranial Aneurysms: Classification Scheme and Management Strategies in 68 Patients. Neurosurgery, 2005, 56, 441-454.	0.6	132
31	Centriolar satellites assemble centrosomal microcephaly proteins to recruit CDK2 and promote centriole duplication. ELife, 2015, 4, .	2.8	118
32	Microcephaly Proteins Wdr62 and Aspm Define a Mother Centriole Complex Regulating Centriole Biogenesis, Apical Complex, and Cell Fate. Neuron, 2016, 92, 813-828.	3.8	116
33	The BabySeq project: implementing genomic sequencing in newborns. BMC Pediatrics, 2018, 18, 225.	0.7	115
34	Recessive gene disruptions in autism spectrum disorder. Nature Genetics, 2019, 51, 1092-1098.	9.4	109
35	Mutations in QARS, Encoding Glutaminyl-tRNA Synthetase, Cause Progressive Microcephaly, Cerebral-Cerebellar Atrophy, and Intractable Seizures. American Journal of Human Genetics, 2014, 94, 547-558.	2.6	106
36	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. Genome Biology, 2014, 15, R53.	13.9	101

#	Article	IF	Citations
37	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. Annals of Neurology, 2019, 86, 821-831.	2.8	96
38	Katanin p80 Regulates Human Cortical Development by Limiting Centriole and Cilia Number. Neuron, 2014, 84, 1240-1257.	3.8	89
39	Prospective, phenotype-driven selection of critically ill neonates for rapid exome sequencing is associated with high diagnostic yield. Genetics in Medicine, 2020, 22, 736-744.	1.1	83
40	Mutation of KCNJ8 in a patient with CantÃº syndrome with unique vascular abnormalities â€“ Support for the role of K(ATP) channels in this condition. European Journal of Medical Genetics, 2013, 56, 678-682.	0.7	79
41	A curated gene list for reporting results of newborn genomic sequencing. Genetics in Medicine, 2017, 19, 809-818.	1.1	79
42	The sensitivity of exome sequencing in identifying pathogenic mutations for LGMD in the United States. Journal of Human Genetics, 2017, 62, 243-252.	1.1	73
43	POMK mutations disrupt muscle development leading to a spectrum of neuromuscular presentations. Human Molecular Genetics, 2014, 23, 5781-5792.	1.4	72
44	Adjusting Head Circumference for Covariates in Autism: Clinical Correlates of a Highly Heritable Continuous Trait. Biological Psychiatry, 2013, 74, 576-584.	0.7	70
45	Recent ultra-rare inherited variants implicate new autism candidate risk genes. Nature Genetics, 2021, 53, 1125-1134.	9.4	68
46	Biallelic mutations in human DCC cause developmental split-brain syndrome. Nature Genetics, 2017, 49, 606-612.	9.4	62
47	Parental interest in genomic sequencing of newborns: enrollment experience from the BabySeq Project. Genetics in Medicine, 2019, 21, 622-630.	1.1	61
48	Unique bioinformatic approach and comprehensive reanalysis improve diagnostic yield of clinical exomes. European Journal of Human Genetics, 2019, 27, 1398-1405.	1.4	60
49	Modest Impact on Risk for Autism Spectrum Disorder of Rare Copy Number Variants at 15<scp>q</scp>11.2, Specifically Breakpoints 1 to 2. Autism Research, 2014, 7, 355-362.	2.1	59
50	Inhibition of Netrin-Mediated Axon Attraction by a Receptor Protein Tyrosine Phosphatase. Science, 2004, 305, 103-106.	6.0	56
51	METTL23, a transcriptional partner of GABPA, is essential for human cognition. Human Molecular Genetics, 2014, 23, 3456-3466.	1.4	47
52	Infant mortality: the contribution of genetic disorders. Journal of Perinatology, 2019, 39, 1611-1619.	0.9	47
53	Discordant results between conventional newborn screening and genomic sequencing in the BabySeq Project. Genetics in Medicine, 2021, 23, 1372-1375.	1.1	47
54	A novel de novo mutation inATP1A3and childhood-onset schizophrenia. Journal of Physical Education and Sports Management, 2016, 2, a001008.	0.5	46

#	Article	IF	Citations
55	Autism and Fragile X Syndrome. Seminars in Neurology, 2014, 34, 258-265.	0.5	43
56	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. American Journal of Human Genetics, 2019, 105, 413-424.	2.6	43
57	DCCmutation update: Congenital mirror movements, isolated agenesis of the corpus callosum, and developmental split brain syndrome. Human Mutation, 2018, 39, 23-39.	1.1	41
58	Integrated genome and transcriptome sequencing identifies a noncoding mutation in the genome replication factor DONSON as the cause of microcephaly-micromelia syndrome. Genome Research, 2017, 27, 1323-1335.	2.4	40
59	Childrenâ€™s rare disease cohorts: an integrative research and clinical genomics initiative. Npj Genomic Medicine, 2020, 5, 29.	1.7	38
60	Psychosocial Effect of Newborn Genomic Sequencing on Families in the BabySeq Project. JAMA Pediatrics, 2021, 175, 1132.	3.3	35
61	BRAT1 mutations present with a spectrum of clinical severity. American Journal of Medical Genetics, Part A, 2016, 170, 2265-2273.	0.7	34
62	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. Genetics in Medicine, 2021, 23, 1028-1040.	1.1	34
63	Psychiatric and Medical Profiles of Autistic Adults in the SPARK Cohort. Journal of Autism and Developmental Disorders, 2020, 50, 3679-3698.	1.7	33
64	Exome sequencing identifies a novel SMCHD1 mutation in facioscapulohumeral muscular dystrophy 2. Neuromuscular Disorders, 2013, 23, 975-980.	0.3	32
65	FLNA genomic rearrangements cause periventricular nodular heterotopia. Neurology, 2012, 78, 269-278.	1.5	23
66	Increased Survival and Partly Preserved Cognition in a Patient WithACO2-Related Disease Secondary to a Novel Variant. Journal of Child Neurology, 2017, 32, 840-845.	0.7	22
67	Overlapping 16p13.11 deletion and gain of copies variations associated with childhood onset psychosis include genes with mechanistic implications for autism associated pathways: Two case reports. American Journal of Medical Genetics, Part A, 2016, 170, 1165-1173.	0.7	16
68	Orthogonal NGS for High Throughput Clinical Diagnostics. Scientific Reports, 2016, 6, 24650.	1.6	14
69	Homozygous deletions implicate non-coding epigenetic marks in Autism spectrum disorder. Scientific Reports, 2020, 10, 14045.	1.6	12
70	Beliefs in vaccine as causes of autism among SPARK cohort caregivers. Vaccine, 2020, 38, 1794-1803.	1.7	12
71	What Name Best Represents Our Specialty? Oral and Maxillofacial Surgeon Versus Oral and Facial Surgeon. Journal of Oral and Maxillofacial Surgery, 2017, 75, 9-20.	0.5	8
72	Reconciling newborn screening and a novel splice variant in BTD associated with partial biotinidase deficiency: a BabySeq Project case report. Journal of Physical Education and Sports Management, 2018, 4, a002873.	0.5	7

#	Article	IF	Citations
73	An Inhibitory Antibody against Dipeptidyl Peptidase IV Improves Glucose Tolerance in Vivo. Journal of Biological Chemistry, 2013, 288, 1307-1316.	1.6	6
74	Rare variant association test in family-based sequencing studies. Briefings in Bioinformatics, 2016, 18, bbw083.	3.2	6
75	Quantifying Downstream Healthcare Utilization in Studies of Genomic Testing. Value in Health, 2020, 23, 559-565.	0.1	6
76	From Sequence Data to Returnable Results: Ethical Issues in Variant Calling and Interpretation. Genetic Testing and Molecular Biomarkers, 2017, 21, 178-183.	0.3	5
77	A phenotypically severe, biochemically â€œsilentâ€•case of HIBCH deficiency in a newborn diagnosed by rapid whole exome sequencing and enzymatic testing. American Journal of Medical Genetics, Part A, 2020, 182, 780-784.	0.7	4
78	Novel variants in KAT6B spectrum of disorders expand our knowledge of clinical manifestations and molecular mechanisms. Molecular Genetics & Genomic Medicine, 2021, 9, e1809.	0.6	4
79	Modified Sensory Testing in Non-verbal Patients Receiving Novel Intrathecal Therapies for Neurological Disorders. Frontiers in Neurology, 2022, 13, 664710.	1.1	2
80	A solid start for gene therapy in Tayâ€“Sachs disease. Nature Medicine, 2022, , .	15.2	2
81	Education Research: A program perspective on learning how to teach. Neurology, 2008, 70, e75-7.	1.5	0
82	OP024: Discovery and therapeutic implications of pathogenic retroelements in neurodegenerative diseases. Genetics in Medicine, 2022, 24, S353-S354.	1.1	0