Timothy W Yu

List of Publications by Year
in descending order

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82 papers	18,794 citations	41344 49 h-index	60623 81 g-index
91 all docs	91 docs citations	91 times ranked	25835 citing authors

#	Article	IF	Citations
1	Modified Sensory Testing in Non-verbal Patients Receiving Novel Intrathecal Therapies for Neurological Disorders. Frontiers in Neurology, 2022, 13, 664710.	2.4	2
2	A solid start for gene therapy in Tayâ€“Sachs disease. Nature Medicine, 2022, , .	30.7	2
3	OP024: Discovery and therapeutic implications of pathogenic retroelements in neurodegenerative diseases. Genetics in Medicine, 2022, 24, S353-S354.	2.4	0
4	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. Genetics in Medicine, 2021, 23, 1028-1040.	2.4	34
5	Discordant results between conventional newborn screening and genomic sequencing in the BabySeq Project. Genetics in Medicine, 2021, 23, 1372-1375.	2.4	47
6	Recent ultra-rare inherited variants implicate new autism candidate risk genes. Nature Genetics, 2021, 53, 1125-1134.	21.4	68
7	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.	2.4	229
8	Psychosocial Effect of Newborn Genomic Sequencing on Families in the BabySeq Project. JAMA Pediatrics, 2021, 175, 1132.	6.2	35
9	Novel variants in KAT6B spectrum of disorders expand our knowledge of clinical manifestations and molecular mechanisms. Molecular Genetics & Genomic Medicine, 2021, 9, e1809.	1.2	4
10	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.	2.4	83
11	Childrenâ€™s rare disease cohorts: an integrative research and clinical genomics initiative. Npj Genomic Medicine, 2020, 5, 29.	3.8	38
12	Homozygous deletions implicate non-coding epigenetic marks in Autism spectrum disorder. Scientific Reports, 2020, 10, 14045.	3.3	12
13	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.	1.2	4
14	Psychiatric and Medical Profiles of Autistic Adults in the SPARK Cohort. Journal of Autism and Developmental Disorders, 2020, 50, 3679-3698.	2.7	33
15	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. Cell, 2020, 180, 568-584.e23.	28.9	1,422
16	Beliefs in vaccine as causes of autism among SPARK cohort caregivers. Vaccine, 2020, 38, 1794-1803.	3.8	12
17	Quantifying Downstream Healthcare Utilization in Studies of Genomic Testing. Value in Health, 2020, 23, 559-565.	0.3	6
18	Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. Npj Genomic Medicine, 2019, 4, 19.	3.8	163

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19	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.	6.2	43
20	Patient-Customized Oligonucleotide Therapy for a Rare Genetic Disease. New England Journal of Medicine, 2019, 381, 1644-1652.	27.0	481
21	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. Annals of Neurology, 2019, 86, 821-831.	5.3	96
22	Infant mortality: the contribution of genetic disorders. Journal of Perinatology, 2019, 39, 1611-1619.	2.0	47
23	Recessive gene disruptions in autism spectrum disorder. Nature Genetics, 2019, 51, 1092-1098.	21.4	109
24	Unique bioinformatic approach and comprehensive reanalysis improve diagnostic yield of clinical exomes. European Journal of Human Genetics, 2019, 27, 1398-1405.	2.8	60
25	Parental interest in genomic sequencing of newborns: enrollment experience from the BabySeq Project. Genetics in Medicine, 2019, 21, 622-630.	2.4	61
26	Interpretation of Genomic Sequencing Results in Healthy and Ill Newborns: Results from the BabySeq Project. American Journal of Human Genetics, 2019, 104, 76-93.	6.2	176
27	DCCmutation update: Congenital mirror movements, isolated agenesis of the corpus callosum, and developmental split brain syndrome. Human Mutation, 2018, 39, 23-39.	2.5	41
28	The BabySeq project: implementing genomic sequencing in newborns. BMC Pediatrics, 2018, 18, 225.	1.7	115
29	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.	1.2	7
30	Newborn Sequencing in Genomic Medicine and Public Health. Pediatrics, 2017, 139, .	2.1	174
31	A curated gene list for reporting results of newborn genomic sequencing. Genetics in Medicine, 2017, 19, 809-818.	2.4	79
32	Biallelic mutations in human DCC cause developmental split-brain syndrome. Nature Genetics, 2017, 49, 606-612.	21.4	62
33	Polygenic transmission disequilibrium confirms that common and rare variation act additively to create risk for autism spectrum disorders. Nature Genetics, 2017, 49, 978-985.	21.4	401
34	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.	5.5	40
35	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.	1.4	22
36	From Sequence Data to Returnable Results: Ethical Issues in Variant Calling and Interpretation. Genetic Testing and Molecular Biomarkers, 2017, 21, 178-183.	0.7	5

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37	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.	4.9	495
38	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.	1.2	8
39	The sensitivity of exome sequencing in identifying pathogenic mutations for LGMD in the United States. Journal of Human Genetics, 2017, 62, 243-252.	2.3	73
40	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.	1.2	16
41	BRAT1 mutations present with a spectrum of clinical severity. American Journal of Medical Genetics, Part A, 2016, 170, 2265-2273.	1.2	34
42	Rare variant association test in family-based sequencing studies. Briefings in Bioinformatics, 2016, 18, bbw083.	6.5	6
43	A novel de novo mutation inATP1A3and childhood-onset schizophrenia. Journal of Physical Education and Sports Management, 2016, 2, a001008.	1.2	46
44	Microcephaly Proteins Wdr62 and Aspm Define a Mother Centriole Complex Regulating Centriole Biogenesis, Apical Complex, and Cell Fate. Neuron, 2016, 92, 813-828.	8.1	116
45	Orthogonal NGS for High Throughput Clinical Diagnostics. Scientific Reports, 2016, 6, 24650.	3.3	14
46	Centriolar satellites assemble centrosomal microcephaly proteins to recruit CDK2 and promote centriole duplication. ELife, 2015, 4, .	6.0	118
47	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.	6.2	225
48	Psychiatric genome-wide association study analyses implicate neuronal, immune and histone pathways. Nature Neuroscience, 2015, 18, 199-209.	14.8	701
49	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. Neuron, 2015, 87, 1215-1233.	8.1	1,219
50	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.	1.3	133
51	METTL23, a transcriptional partner of GABPA, is essential for human cognition. Human Molecular Genetics, 2014, 23, 3456-3466.	2.9	47
52	Katanin p80 Regulates Human Cortical Development by Limiting Centriole and Cilia Number. Neuron, 2014, 84, 1240-1257.	8.1	89
53	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.	9.6	101
54	POMK mutations disrupt muscle development leading to a spectrum of neuromuscular presentations. Human Molecular Genetics, 2014, 23, 5781-5792.	2.9	72

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55	Autism and Fragile X Syndrome. Seminars in Neurology, 2014, 34, 258-265.	1.4	43
56	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.	6.2	106
57	Synaptic, transcriptional and chromatin genes disrupted in autism. Nature, 2014, 515, 209-215.	27.8	2,254
58	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.	3.8	59
59	The ion channel TRPV1 regulates the activation and proinflammatory properties of CD4+ T cells. Nature Immunology, 2014, 15, 1055-1063.	14.5	193
60	Somatic Mutations in Cerebral Cortical Malformations. New England Journal of Medicine, 2014, 371, 733-743.	27.0	326
61	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.	6.2	147
62	Adjusting Head Circumference for Covariates in Autism: Clinical Correlates of a Highly Heritable Continuous Trait. Biological Psychiatry, 2013, 74, 576-584.	1.3	70
63	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. Nature Genetics, 2013, 45, 984-994.	21.4	2,067
64	An Inhibitory Antibody against Dipeptidyl Peptidase IV Improves Glucose Tolerance in Vivo. Journal of Biological Chemistry, 2013, 288, 1307-1316.	3.4	6
65	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.	1.3	79
66	Exome sequencing identifies a novel SMCHD1 mutation in facioscapulohumeral muscular dystrophy 2. Neuromuscular Disorders, 2013, 23, 975-980.	0.6	32
67	Using Whole-Exome Sequencing to Identify Inherited Causes of Autism. Neuron, 2013, 77, 259-273.	8.1	383
68	FLNA genomic rearrangements cause periventricular nodular heterotopia. Neurology, 2012, 78, 269-278.	1.1	23
69	Whole-Exome Sequencing and Homozygosity Analysis Implicate Depolarization-Regulated Neuronal Genes in Autism. PLoS Genetics, 2012, 8, e1002635.	3.5	164
70	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.	6.2	167
71	Common genetic variants, acting additively, are a major source of risk for autism. Molecular Autism, 2012, 3, 9.	4.9	357
72	Multiple Recurrent De Novo CNVs, Including Duplications of the 7q11.23 Williams Syndrome Region, Are Strongly Associated with Autism. Neuron, 2011, 70, 863-885.	8.1	1,146

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73	Mutations in WDR62, encoding a centrosome-associated protein, cause microcephaly with simplified gyri and abnormal cortical architecture. Nature Genetics, 2010, 42, 1015-1020.	21.4	259
74	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.	7.1	372
75	Education Research: A program perspective on learning how to teach. Neurology, 2008, 70, e75-7.	1.1	0
76	Thrombotic Intracranial Aneurysms: Classification Scheme and Management Strategies in 68 Patients. Neurosurgery, 2005, 56, 441-454.	1.1	132
77	Inhibition of Netrin-Mediated Axon Attraction by a Receptor Protein Tyrosine Phosphatase. Science, 2004, 305, 103-106.	12.6	56
78	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.	8.1	216
79	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.	14.8	144
80	C. elegans Slit Acts in Midline, Dorsal-Ventral, and Anterior-Posterior Guidance via the SAX-3/Robo Receptor. Neuron, 2001, 32, 25-38.	8.1	209
81	Dynamic regulation of axon guidance. Nature Neuroscience, 2001, 4, 1169-1176.	14.8	294
82	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.	3.6	1,744