

# Timothy W Yu

## List of Publications by Year in descending order

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Version: 2024-02-01

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	Modified Sensory Testing in Non-verbal Patients Receiving Novel Intrathecal Therapies for Neurological Disorders. <i>Frontiers in Neurology</i> , 2022, 13, 664710.	1.1	2
2	A solid start for gene therapy in Tayâ€“Sachs disease. <i>Nature Medicine</i> , 2022, , .	15.2	2
3	OPO24: Discovery and therapeutic implications of pathogenic retroelements in neurodegenerative diseases. <i>Genetics in Medicine</i> , 2022, 24, S353-S354.	1.1	0
4	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. <i>Genetics in Medicine</i> , 2021, 23, 1028-1040.	1.1	34
5	Discordant results between conventional newborn screening and genomic sequencing in the BabySeq Project. <i>Genetics in Medicine</i> , 2021, 23, 1372-1375.	1.1	47
6	Recent ultra-rare inherited variants implicate new autism candidate risk genes. <i>Nature Genetics</i> , 2021, 53, 1125-1134.	9.4	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). <i>Genetics in Medicine</i> , 2021, 23, 2029-2037.	1.1	229
8	Psychosocial Effect of Newborn Genomic Sequencing on Families in the BabySeq Project. <i>JAMA Pediatrics</i> , 2021, 175, 1132.	3.3	35
9	Novel variants in <i>KAT6B</i> spectrum of disorders expand our knowledge of clinical manifestations and molecular mechanisms. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2021, 9, e1809.	0.6	4
10	Prospective, phenotype-driven selection of critically ill neonates for rapid exome sequencing is associated with high diagnostic yield. <i>Genetics in Medicine</i> , 2020, 22, 736-744.	1.1	83
11	Childrenâ€™s rare disease cohorts: an integrative research and clinical genomics initiative. <i>Npj Genomic Medicine</i> , 2020, 5, 29.	1.7	38
12	Homozygous deletions implicate non-coding epigenetic marks in Autism spectrum disorder. <i>Scientific Reports</i> , 2020, 10, 14045.	1.6	12
13	A phenotypically severe, biochemically â€œsilentâ€–case of HIBCH deficiency in a newborn diagnosed by rapid whole exome sequencing and enzymatic testing. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 780-784.	0.7	4
14	Psychiatric and Medical Profiles of Autistic Adults in the SPARK Cohort. <i>Journal of Autism and Developmental Disorders</i> , 2020, 50, 3679-3698.	1.7	33
15	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. <i>Cell</i> , 2020, 180, 568-584.e23.	13.5	1,422
16	Beliefs in vaccine as causes of autism among SPARK cohort caregivers. <i>Vaccine</i> , 2020, 38, 1794-1803.	1.7	12
17	Quantifying Downstream Healthcare Utilization in Studies of Genomic Testing. <i>Value in Health</i> , 2020, 23, 559-565.	0.1	6
18	Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. <i>Npj Genomic Medicine</i> , 2019, 4, 19.	1.7	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.	2.6	43
20	Patient-Customized Oligonucleotide Therapy for a Rare Genetic Disease. New England Journal of Medicine, 2019, 381, 1644-1652.	13.9	481
21	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. Annals of Neurology, 2019, 86, 821-831.	2.8	96
22	Infant mortality: the contribution of genetic disorders. Journal of Perinatology, 2019, 39, 1611-1619.	0.9	47
23	Recessive gene disruptions in autism spectrum disorder. Nature Genetics, 2019, 51, 1092-1098.	9.4	109
24	Unique bioinformatic approach and comprehensive reanalysis improve diagnostic yield of clinical exomes. European Journal of Human Genetics, 2019, 27, 1398-1405.	1.4	60
25	Parental interest in genomic sequencing of newborns: enrollment experience from the BabySeq Project. Genetics in Medicine, 2019, 21, 622-630.	1.1	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.	2.6	176
27	<i>DCC</i> mutation update: Congenital mirror movements, isolated agenesis of the corpus callosum, and developmental split brain syndrome. Human Mutation, 2018, 39, 23-39.	1.1	41
28	The BabySeq project: implementing genomic sequencing in newborns. BMC Pediatrics, 2018, 18, 225.	0.7	115
29	Reconciling newborn screening and a novel splice variant in <i>BTD</i> associated with partial biotinidase deficiency: a BabySeq Project case report. Journal of Physical Education and Sports Management, 2018, 4, a002873.	0.5	7
30	Newborn Sequencing in Genomic Medicine and Public Health. Pediatrics, 2017, 139, .	1.0	174
31	A curated gene list for reporting results of newborn genomic sequencing. Genetics in Medicine, 2017, 19, 809-818.	1.1	79
32	Biallelic mutations in human DCC cause developmental split-brain syndrome. Nature Genetics, 2017, 49, 606-612.	9.4	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.	9.4	401
34	Integrated genome and transcriptome sequencing identifies a noncoding mutation in the genome replication factor <i>DONSON</i> as the cause of microcephaly-micromelia syndrome. Genome Research, 2017, 27, 1323-1335.	2.4	40
35	Increased Survival and Partly Preserved Cognition in a Patient With <i>ACO2</i> -Related Disease Secondary to a Novel Variant. Journal of Child Neurology, 2017, 32, 840-845.	0.7	22
36	From Sequence Data to Returnable Results: Ethical Issues in Variant Calling and Interpretation. Genetic Testing and Molecular Biomarkers, 2017, 21, 178-183.	0.3	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. <i>Molecular Autism</i> , 2017, 8, 21.	2.6	495
38	What Name Best Represents Our Specialty? Oral and Maxillofacial Surgeon Versus Oral and Facial Surgeon. <i>Journal of Oral and Maxillofacial Surgery</i> , 2017, 75, 9-20.	0.5	8
39	The sensitivity of exome sequencing in identifying pathogenic mutations for LGMD in the United States. <i>Journal of Human Genetics</i> , 2017, 62, 243-252.	1.1	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. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1165-1173.	0.7	16
41	<i>BRAT1</i> mutations present with a spectrum of clinical severity. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2265-2273.	0.7	34
42	Rare variant association test in family-based sequencing studies. <i>Briefings in Bioinformatics</i> , 2016, 18, bbw083.	3.2	6
43	A novel de novo mutation in <i>ATP1A3</i> and childhood-onset schizophrenia. <i>Journal of Physical Education and Sports Management</i> , 2016, 2, a001008.	0.5	46
44	Microcephaly Proteins Wdr62 and Aspm Define a Mother Centriole Complex Regulating Centriole Biogenesis, Apical Complex, and Cell Fate. <i>Neuron</i> , 2016, 92, 813-828.	3.8	116
45	Orthogonal NGS for High Throughput Clinical Diagnostics. <i>Scientific Reports</i> , 2016, 6, 24650.	1.6	14
46	Centriolar satellites assemble centrosomal microcephaly proteins to recruit CDK2 and promote centriole duplication. <i>ELife</i> , 2015, 4, .	2.8	118
47	Joint Analysis of Psychiatric Disorders Increases Accuracy of Risk Prediction for Schizophrenia, Bipolar Disorder, and Major Depressive Disorder. <i>American Journal of Human Genetics</i> , 2015, 96, 283-294.	2.6	225
48	Psychiatric genome-wide association study analyses implicate neuronal, immune and histone pathways. <i>Nature Neuroscience</i> , 2015, 18, 199-209.	7.1	701
49	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. <i>Neuron</i> , 2015, 87, 1215-1233.	3.8	1,219
50	A Genome-wide Association Study of Autism Using the Simons Simplex Collection: Does Reducing Phenotypic Heterogeneity in Autism Increase Genetic Homogeneity?. <i>Biological Psychiatry</i> , 2015, 77, 775-784.	0.7	133
51	<i>METTL23</i> , a transcriptional partner of <i>GABPA</i> , is essential for human cognition. <i>Human Molecular Genetics</i> , 2014, 23, 3456-3466.	1.4	47
52	Katanin p80 Regulates Human Cortical Development by Limiting Centriole and Cilia Number. <i>Neuron</i> , 2014, 84, 1240-1257.	3.8	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. <i>Genome Biology</i> , 2014, 15, R53.	13.9	101
54	<i>POMK</i> mutations disrupt muscle development leading to a spectrum of neuromuscular presentations. <i>Human Molecular Genetics</i> , 2014, 23, 5781-5792.	1.4	72

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55	Autism and Fragile X Syndrome. <i>Seminars in Neurology</i> , 2014, 34, 258-265.	0.5	43
56	Mutations in QARS, Encoding Glutaminyl-tRNA Synthetase, Cause Progressive Microcephaly, Cerebral-Cerebellar Atrophy, and Intractable Seizures. <i>American Journal of Human Genetics</i> , 2014, 94, 547-558.	2.6	106
57	Synaptic, transcriptional and chromatin genes disrupted in autism. <i>Nature</i> , 2014, 515, 209-215.	13.7	2,254
58	Modest Impact on Risk for Autism Spectrum Disorder of Rare Copy Number Variants at 15q11.2, Specifically Breakpoints 1 to 2. <i>Autism Research</i> , 2014, 7, 355-362.	2.1	59
59	The ion channel TRPV1 regulates the activation and proinflammatory properties of CD4+ T cells. <i>Nature Immunology</i> , 2014, 15, 1055-1063.	7.0	193
60	Somatic Mutations in Cerebral Cortical Malformations. <i>New England Journal of Medicine</i> , 2014, 371, 733-743.	13.9	326
61	Identification of KLHL41 Mutations Implicates BTB-Kelch-Mediated Ubiquitination as an Alternate Pathway to Myofibrillar Disruption in Nemaline Myopathy. <i>American Journal of Human Genetics</i> , 2013, 93, 1108-1117.	2.6	147
62	Adjusting Head Circumference for Covariates in Autism: Clinical Correlates of a Highly Heritable Continuous Trait. <i>Biological Psychiatry</i> , 2013, 74, 576-584.	0.7	70
63	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. <i>Nature Genetics</i> , 2013, 45, 984-994.	9.4	2,067
64	An Inhibitory Antibody against Dipeptidyl Peptidase IV Improves Glucose Tolerance in Vivo. <i>Journal of Biological Chemistry</i> , 2013, 288, 1307-1316.	1.6	6
65	Mutation of KCNJ8 in a patient with Cantu syndrome with unique vascular abnormalities – Support for the role of K(ATP) channels in this condition. <i>European Journal of Medical Genetics</i> , 2013, 56, 678-682.	0.7	79
66	Exome sequencing identifies a novel SMCHD1 mutation in facioscapulohumeral muscular dystrophy 2. <i>Neuromuscular Disorders</i> , 2013, 23, 975-980.	0.3	32
67	Using Whole-Exome Sequencing to Identify Inherited Causes of Autism. <i>Neuron</i> , 2013, 77, 259-273.	3.8	383
68	FLNA genomic rearrangements cause periventricular nodular heterotopia. <i>Neurology</i> , 2012, 78, 269-278.	1.5	23
69	Whole-Exome Sequencing and Homozygosity Analysis Implicate Depolarization-Regulated Neuronal Genes in Autism. <i>PLoS Genetics</i> , 2012, 8, e1002635.	1.5	164
70	Exome Sequencing and Functional Validation in Zebrafish Identify GTDC2 Mutations as a Cause of Walker-Warburg Syndrome. <i>American Journal of Human Genetics</i> , 2012, 91, 541-547.	2.6	167
71	Common genetic variants, acting additively, are a major source of risk for autism. <i>Molecular Autism</i> , 2012, 3, 9.	2.6	357
72	Multiple Recurrent De Novo CNVs, Including Duplications of the 7q11.23 Williams Syndrome Region, Are Strongly Associated with Autism. <i>Neuron</i> , 2011, 70, 863-885.	3.8	1,146

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73	Mutations in WDR62, encoding a centrosome-associated protein, cause microcephaly with simplified gyri and abnormal cortical architecture. <i>Nature Genetics</i> , 2010, 42, 1015-1020.	9.4	259
74	A proprotein convertase subtilisin/kexin type 9 neutralizing antibody reduces serum cholesterol in mice and nonhuman primates. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2009, 106, 9820-9825.	3.3	372
75	Education Research: A program perspective on learning how to teach. <i>Neurology</i> , 2008, 70, e75-7.	1.5	0
76	Thrombotic Intracranial Aneurysms: Classification Scheme and Management Strategies in 68 Patients. <i>Neurosurgery</i> , 2005, 56, 441-454.	0.6	132
77	Inhibition of Netrin-Mediated Axon Attraction by a Receptor Protein Tyrosine Phosphatase. <i>Science</i> , 2004, 305, 103-106.	6.0	56
78	The Netrin Receptor UNC-40/DCC Stimulates Axon Attraction and Outgrowth through Enabled and, in Parallel, Rac and UNC-115/AbLIM. <i>Neuron</i> , 2003, 37, 53-65.	3.8	216
79	Shared receptors in axon guidance: SAX-3/Robo signals via UNC-34/Enabled and a Netrin-independent UNC-40/DCC function. <i>Nature Neuroscience</i> , 2002, 5, 1147-1154.	7.1	144
80	<i>C. elegans</i> Slit Acts in Midline, Dorsal-Ventral, and Anterior-Posterior Guidance via the SAX-3/Robo Receptor. <i>Neuron</i> , 2001, 32, 25-38.	3.8	209
81	Dynamic regulation of axon guidance. <i>Nature Neuroscience</i> , 2001, 4, 1169-1176.	7.1	294
82	Dentate Granule Cell Neurogenesis Is Increased by Seizures and Contributes to Aberrant Network Reorganization in the Adult Rat Hippocampus. <i>Journal of Neuroscience</i> , 1997, 17, 3727-3738.	1.7	1,744