

# Hung-Chun Yu

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/4780092/publications.pdf>

Version: 2024-02-01

20  
papers

1,240  
citations

687363

13  
h-index

752698

20  
g-index

24  
all docs

24  
docs citations

24  
times ranked

3857  
citing authors

#	ARTICLE	IF	CITATIONS
1	Whole-exome sequencing and adrenocorticotrophic hormone therapy in individuals with infantile spasms. <i>Developmental Medicine and Child Neurology</i> , 2022, 64, 633-640.	2.1	4
2	Genome-wide copy number variations in a large cohort of bantu African children. <i>BMC Medical Genomics</i> , 2021, 14, 129.	1.5	6
3	An unsupervised learning approach to identify novel signatures of health and disease from multimodal data. <i>Genome Medicine</i> , 2020, 12, 7.	8.2	27
4	Abnormal expression of GABAA receptor sub-units and hypomotility upon loss of <i>gabral1</i> in zebrafish. <i>Biology Open</i> , 2020, 9, .	1.2	12
5	Precision medicine integrating whole-genome sequencing, comprehensive metabolomics, and advanced imaging. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020, 117, 3053-3062.	7.1	85
6	Response to Whiffin et al.. <i>American Journal of Human Genetics</i> , 2019, 104, 186.	6.2	1
7	The human noncoding genome defined by genetic diversity. <i>Nature Genetics</i> , 2018, 50, 333-337.	21.4	137
8	Identification of Misclassified ClinVar Variants via Disease Population Prevalence. <i>American Journal of Human Genetics</i> , 2018, 102, 609-619.	6.2	117
9	Mutations in PMPCB Encoding the Catalytic Subunit of the Mitochondrial Presequence Protease Cause Neurodegeneration in Early Childhood. <i>American Journal of Human Genetics</i> , 2018, 102, 557-573.	6.2	69
10	Precision medicine screening using whole-genome sequencing and advanced imaging to identify disease risk in adults. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, 3686-3691.	7.1	76
11	Whole-genome sequencing identifies common-to-rare variants associated with human blood metabolites. <i>Nature Genetics</i> , 2017, 49, 568-578.	21.4	341
12	Mutations in THAP11 cause an inborn error of cobalamin metabolism and developmental abnormalities. <i>Human Molecular Genetics</i> , 2017, 26, 2838-2849.	2.9	47
13	X-Linked Cobalamin Disorder ( HCFC1 ) Mimicking Nonketotic Hyperglycinemia With Increased Both Cerebrospinal Fluid Glycine and Methylmalonic Acid. <i>Pediatric Neurology</i> , 2017, 71, 65-69.	2.1	11
14	Discovery of a potentially deleterious variant in <i>TMEM87B</i> in a patient with a hemizygous 2q13 microdeletion suggests a recessive condition characterized by congenital heart disease and restrictive cardiomyopathy. <i>Journal of Physical Education and Sports Management</i> , 2016, 2, a000844.	1.2	18
15	Compound heterozygosity for a frame shift mutation and a likely pathogenic sequence variant in the planar cell polarity-ciliogenesis gene <i>WDPCP</i> in a girl with polysyndactyly, coarctation of the aorta, and tongue hamartomas. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 421-427.	1.2	26
16	Mutations in the mitochondrial cysteinyl-tRNA synthase gene, <i>CARS2</i> , lead to a severe epileptic encephalopathy and complex movement disorder. <i>Journal of Medical Genetics</i> , 2015, 52, 532-540.	3.2	62
17	An individual with blepharophimosis-ptosis-epicanthus inversus syndrome (BPES) and additional features expands the phenotype associated with mutations in <i>KAT6B</i> . <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 950-957.	1.2	33
18	Evaluating Familial Essential Tremor with Novel Genetic Approaches: Is it a Genotyping or Phenotyping Issue?. <i>Tremor and Other Hyperkinetic Movements</i> , 2014, 4, 258.	2.0	4

#	ARTICLE	IF	CITATIONS
19	An X-Linked Cobalamin Disorder Caused by Mutations in Transcriptional Coregulator HCFC1. American Journal of Human Genetics, 2013, 93, 506-514.	6.2	110
20	Targeted discovery of novel human exons by comparative genomics. Genome Research, 2007, 17, 1763-1773.	5.5	42