Hung-Chun Yu

List of Publications by Year
in descending order

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

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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-genome sequencing identifies common-to-rare variants associated with human blood metabolites. Nature Genetics, 2017, 49, 568-578.	21.4	341
2	The human noncoding genome defined by genetic diversity. Nature Genetics, 2018, 50, 333-337.	21.4	137
3	Identification of Misclassified ClinVar Variants via Disease Population Prevalence. American Journal of Human Genetics, 2018, 102, 609-619.	6.2	117
4	An X-Linked Cobalamin Disorder Caused by Mutations in Transcriptional Coregulator HCFC1. American Journal of Human Genetics, 2013, 93, 506-514.	6.2	110
5	Precision medicine integrating whole-genome sequencing, comprehensive metabolomics, and advanced imaging. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 3053-3062.	7.1	85
6	Precision medicine screening using whole-genome sequencing and advanced imaging to identify disease risk in adults. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 3686-3691.	7.1	76
7	Mutations in PMPCB Encoding the Catalytic Subunit of the Mitochondrial Presequence Protease Cause Neurodegeneration in Early Childhood. American Journal of Human Genetics, 2018, 102, 557-573.	6.2	69
8	Mutations in the mitochondrial cysteinyl-tRNA synthase gene,CARS2,lead to a severe epileptic encephalopathy and complex movement disorder. Journal of Medical Genetics, 2015, 52, 532-540.	3.2	62
9	Mutations in THAP11 cause an inborn error of cobalamin metabolism and developmental abnormalities. Human Molecular Genetics, 2017, 26, 2838-2849.	2.9	47
10	Targeted discovery of novel human exons by comparative genomics. Genome Research, 2007, 17, 1763-1773.	5.5	42
11	An individual with blepharophimosisâ€“ptosisâ€“epicanthus inversus syndrome (BPES) and additional features expands the phenotype associated with mutations in KAT6B. American Journal of Medical Genetics, Part A, 2014, 164, 950-957.	1.2	33
12	An unsupervised learning approach to identify novel signatures of health and disease from multimodal data. Genome Medicine, 2020, 12, 7.	8.2	27
13	Compound heterozygosity for a frame shift mutation and a likely pathogenic sequence variant in the planar cell polarityâ€“ciliogenesis gene WDPCP in a girl with polysyndactyly, coarctation of the aorta, and tongue hamartomas. American Journal of Medical Genetics, Part A, 2015, 167, 421-427.	1.2	26
14	Discovery of a potentially deleterious variant in TMEM87B in a patient with a hemizygous 2q13 microdeletion suggests a recessive condition characterized by congenital heart disease and restrictive cardiomyopathy. Journal of Physical Education and Sports Management, 2016, 2, a000844.	1.2	18
15	Abnormal expression of GABAA receptor sub-units and hypomotility upon loss of gabra1 in zebrafish. Biology Open, 2020, 9, .	1.2	12
16	X-Linked Cobalamin Disorder (HCFC1) Mimicking Nonketotic Hyperglycinemia With Increased Both Cerebrospinal Fluid Glycine and Methylmalonic Acid. Pediatric Neurology, 2017, 71, 65-69.	2.1	11
17	Genome-wide copy number variations in a large cohort of bantu African children. BMC Medical Genomics, 2021, 14, 129.	1.5	6
18	Evaluating Familial Essential Tremor with Novel Genetic Approaches: Is it a Genotyping or Phenotyping Issue?. Tremor and Other Hyperkinetic Movements, 2014, 4, 258.	2.0	4

#	Article	IF	Citations
19	Wholeâ€exome sequencing and adrenocorticotropic hormone therapy in individuals with infantile spasms. Developmental Medicine and Child Neurology, 2022, 64, 633-640.	2.1	4
20	Response to Whiffin etÂal.. American Journal of Human Genetics, 2019, 104, 186.	6.2	1