Colby Chiang

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/4365384/publications.pdf

Version: 2024-02-01

26 papers	4,836 citations	331670 21 h-index	552781 26 g-index
34 all docs	34 docs citations	34 times ranked	10610 citing authors

#	Article	IF	Citations
1	Association of structural variation with cardiometabolic traits in Finns. American Journal of Human Genetics, 2021, 108, 583-596.	6.2	22
2	Structural variants are a major source of gene expression differences in humans and often affect multiple nearby genes. Genome Research, 2021, 31, 2249-2257.	5.5	48
3	Mapping and characterization of structural variation in 17,795 human genomes. Nature, 2020, 583, 83-89.	27.8	194
4	svtools: population-scale analysis of structural variation. Bioinformatics, 2019, 35, 4782-4787.	4.1	51
5	Identification of Drivers of Aneuploidy in Breast Tumors. Cell Reports, 2018, 23, 2758-2769.	6.4	57
6	Cover Image, Volume 173A, Number 2, February 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
7	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248.	21.4	131
8	Potential molecular consequences of transgene integration: The R6/2 mouse example. Scientific Reports, 2017, 7, 41120.	3.3	14
9	The impact of structural variation on human gene expression. Nature Genetics, 2017, 49, 692-699.	21.4	334
10	The impact of rare variation on gene expression across tissues. Nature, 2017, 550, 239-243.	27.8	229
11	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45.	21.4	251
12	Implication of LRRC4C and DPP6 in neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2017, 173, 395-406.	1.2	40
13	Mutations in DCHS1 cause mitral valve prolapse. Nature, 2015, 525, 109-113.	27.8	150
14	SpeedSeq: ultra-fast personal genome analysis and interpretation. Nature Methods, 2015, 12, 966-968.	19.0	515
15	The genome of the vervet (Chlorocebus aethiops sabaeus). Genome Research, 2015, 25, 1921-1933.	5.5	114
16	LUMPY: a probabilistic framework for structural variant discovery. Genome Biology, 2014, 15, R84.	9.6	1,199
17	Lack of association of rare functional variants in TSC1/TSC2 genes with autism spectrum disorder. Molecular Autism, 2013, 4, 5.	4.9	16
18	Haploinsufficiency of KDM6A is associated with severe psychomotor retardation, global growth restriction, seizures and cleft palate. Human Genetics, 2013, 132, 537-552.	3.8	60

#	Article	IF	Citations
19	Molecular Analysis of a Deletion Hotspot in the NRXN1 Region Reveals the Involvement of Short Inverted Repeats in Deletion CNVs. American Journal of Human Genetics, 2013, 92, 375-386.	6.2	42
20	Exonic Deletions in AUTS2 Cause a Syndromic Form of Intellectual Disability and Suggest a Critical Role for the C Terminus. American Journal of Human Genetics, 2013, 92, 210-220.	6.2	135
21	Highly Penetrant Alterations of a Critical Region Including BDNF in Human Psychopathology and Obesity. Archives of General Psychiatry, 2012, 69, 1238.	12.3	22
22	Complex reorganization and predominant non-homologous repair following chromosomal breakage in karyotypically balanced germline rearrangements and transgenic integration. Nature Genetics, 2012, 44, 390-397.	21.4	229
23	Sequencing Chromosomal Abnormalities Reveals Neurodevelopmental Loci that Confer Risk across Diagnostic Boundaries. Cell, 2012, 149, 525-537.	28.9	534
24	Disruption of a Large Intergenic Noncoding RNA in Subjects with Neurodevelopmental Disabilities. American Journal of Human Genetics, 2012, 91, 1128-1134.	6.2	61
25	Next-Generation Sequencing Strategies Enable Routine Detection of Balanced Chromosome Rearrangements for Clinical Diagnostics and Genetic Research. American Journal of Human Genetics, 2011, 88, 469-481.	6.2	154
26	Assessment of 2q23.1 Microdeletion Syndrome Implicates MBD5 as a Single Causal Locus of Intellectual Disability, Epilepsy, and Autism Spectrum Disorder. American Journal of Human Genetics, 2011, 89, 551-563.	6.2	195