Colby Chiang

List of Publications by Year
in descending order

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

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26 papers	4,836 citations	331538 21 h-index	552653 26 g-index
34 all docs	34 docs citations	34 times ranked	10610 citing authors

#	Article	IF	Citations
1	LUMPY: a probabilistic framework for structural variant discovery. Genome Biology, 2014, 15, R84.	13.9	1,199
2	Sequencing Chromosomal Abnormalities Reveals Neurodevelopmental Loci that Confer Risk across Diagnostic Boundaries. Cell, 2012, 149, 525-537.	13.5	534
3	SpeedSeq: ultra-fast personal genome analysis and interpretation. Nature Methods, 2015, 12, 966-968.	9.0	515
4	The impact of structural variation on human gene expression. Nature Genetics, 2017, 49, 692-699.	9.4	334
5	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45.	9.4	251
6	Complex reorganization and predominant non-homologous repair following chromosomal breakage in karyotypically balanced germline rearrangements and transgenic integration. Nature Genetics, 2012, 44, 390-397.	9.4	229
7	The impact of rare variation on gene expression across tissues. Nature, 2017, 550, 239-243.	13.7	229
8	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.	2.6	195
9	Mapping and characterization of structural variation in 17,795 human genomes. Nature, 2020, 583, 83-89.	13.7	194
10	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.	2.6	154
11	Mutations in DCHS1 cause mitral valve prolapse. Nature, 2015, 525, 109-113.	13.7	150
12	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.	2.6	135
13	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248.	9.4	131
14	The genome of the vervet (Chlorocebus aethiops sabaeus). Genome Research, 2015, 25, 1921-1933.	2.4	114
15	Disruption of a Large Intergenic Noncoding RNA in Subjects with Neurodevelopmental Disabilities. American Journal of Human Genetics, 2012, 91, 1128-1134.	2.6	61
16	Haploinsufficiency of KDM6A is associated with severe psychomotor retardation, global growth restriction, seizures and cleft palate. Human Genetics, 2013, 132, 537-552.	1.8	60
17	Identification of Drivers of Aneuploidy in Breast Tumors. Cell Reports, 2018, 23, 2758-2769.	2.9	57
18	svtools: population-scale analysis of structural variation. Bioinformatics, 2019, 35, 4782-4787.	1.8	51

#	Article	IF	Citations
19	Structural variants are a major source of gene expression differences in humans and often affect multiple nearby genes. Genome Research, 2021, 31, 2249-2257.	2.4	48
20	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.	2.6	42
21	Implication of LRRC4C and DPP6 in neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2017, 173, 395-406.	0.7	40
22	Highly Penetrant Alterations of a Critical Region Including BDNF in Human Psychopathology and Obesity. Archives of General Psychiatry, 2012, 69, 1238.	13.8	22
23	Association of structural variation with cardiometabolic traits in Finns. American Journal of Human Genetics, 2021, 108, 583-596.	2.6	22
24	Lack of association of rare functional variants in TSC1/TSC2 genes with autism spectrum disorder. Molecular Autism, 2013, 4, 5.	2.6	16
25	Potential molecular consequences of transgene integration: The R6/2 mouse example. Scientific Reports, 2017, 7, 41120.	1.6	14
26	Cover Image, Volume 173A, Number 2, February 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	0.7	0