

# 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	LUMPY: a probabilistic framework for structural variant discovery. <i>Genome Biology</i> , 2014, 15, R84.	9.6	1,199
2	Sequencing Chromosomal Abnormalities Reveals Neurodevelopmental Loci that Confer Risk across Diagnostic Boundaries. <i>Cell</i> , 2012, 149, 525-537.	28.9	534
3	SpeedSeq: ultra-fast personal genome analysis and interpretation. <i>Nature Methods</i> , 2015, 12, 966-968.	19.0	515
4	The impact of structural variation on human gene expression. <i>Nature Genetics</i> , 2017, 49, 692-699.	21.4	334
5	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. <i>Nature Genetics</i> , 2017, 49, 36-45.	21.4	251
6	Complex reorganization and predominant non-homologous repair following chromosomal breakage in karyotypically balanced germline rearrangements and transgenic integration. <i>Nature Genetics</i> , 2012, 44, 390-397.	21.4	229
7	The impact of rare variation on gene expression across tissues. <i>Nature</i> , 2017, 550, 239-243.	27.8	229
8	Assessment of 2q23.1 Microdeletion Syndrome Implicates MBD5 as a Single Causal Locus of Intellectual Disability, Epilepsy, and Autism Spectrum Disorder. <i>American Journal of Human Genetics</i> , 2011, 89, 551-563.	6.2	195
9	Mapping and characterization of structural variation in 17,795 human genomes. <i>Nature</i> , 2020, 583, 83-89.	27.8	194
10	Next-Generation Sequencing Strategies Enable Routine Detection of Balanced Chromosome Rearrangements for Clinical Diagnostics and Genetic Research. <i>American Journal of Human Genetics</i> , 2011, 88, 469-481.	6.2	154
11	Mutations in DCHS1 cause mitral valve prolapse. <i>Nature</i> , 2015, 525, 109-113.	27.8	150
12	Exonic Deletions in AUTS2 Cause a Syndromic Form of Intellectual Disability and Suggest a Critical Role for the C Terminus. <i>American Journal of Human Genetics</i> , 2013, 92, 210-220.	6.2	135
13	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. <i>Nature Genetics</i> , 2017, 49, 238-248.	21.4	131
14	The genome of the vervet ( <i>Chlorocebus aethiops sabaeus</i> ). <i>Genome Research</i> , 2015, 25, 1921-1933.	5.5	114
15	Disruption of a Large Intergenic Noncoding RNA in Subjects with Neurodevelopmental Disabilities. <i>American Journal of Human Genetics</i> , 2012, 91, 1128-1134.	6.2	61
16	Haploinsufficiency of KDM6A is associated with severe psychomotor retardation, global growth restriction, seizures and cleft palate. <i>Human Genetics</i> , 2013, 132, 537-552.	3.8	60
17	Identification of Drivers of Aneuploidy in Breast Tumors. <i>Cell Reports</i> , 2018, 23, 2758-2769.	6.4	57
18	svtools: population-scale analysis of structural variation. <i>Bioinformatics</i> , 2019, 35, 4782-4787.	4.1	51

#	ARTICLE	IF	CITATIONS
19	Structural variants are a major source of gene expression differences in humans and often affect multiple nearby genes. <i>Genome Research</i> , 2021, 31, 2249-2257.	5.5	48
20	Molecular Analysis of a Deletion Hotspot in the NRXN1 Region Reveals the Involvement of Short Inverted Repeats in Deletion CNVs. <i>American Journal of Human Genetics</i> , 2013, 92, 375-386.	6.2	42
21	Implication of <i>LRRC4C</i> and <i>DPP6</i> in neurodevelopmental disorders. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 395-406.	1.2	40
22	Highly Penetrant Alterations of a Critical Region Including BDNF in Human Psychopathology and Obesity. <i>Archives of General Psychiatry</i> , 2012, 69, 1238.	12.3	22
23	Association of structural variation with cardiometabolic traits in Finns. <i>American Journal of Human Genetics</i> , 2021, 108, 583-596.	6.2	22
24	Lack of association of rare functional variants in TSC1/TSC2 genes with autism spectrum disorder. <i>Molecular Autism</i> , 2013, 4, 5.	4.9	16
25	Potential molecular consequences of transgene integration: The R6/2 mouse example. <i>Scientific Reports</i> , 2017, 7, 41120.	3.3	14
26	Cover Image, Volume 173A, Number 2, February 2017. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, i.	1.2	0