Ryan L Collins, Ab

List of Publications by Year
in descending order

35 papers	10,518 citations	249298 26 h-index	406436 35 g-index
52 all docs	52 docs citations	52 times ranked	23030 citing authors

#	Article	IF	Citations
1	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	1.1	44
2	Mutations causing Lopes-Maciel-Rodan syndrome are huntingtin hypomorphs. Human Molecular Genetics, 2021, 30, 135-148.	1.4	24
3	Familial thrombocytopenia due to a complex structural variant resulting in a WAC-ANKRD26 fusion transcript. Journal of Experimental Medicine, 2021, 218, .	4.2	20
4	Genome-wide enhancer maps link risk variants to disease genes. Nature, 2021, 593, 238-243.	13.7	332
5	Expectations and blind spots for structural variation detection from long-read assemblies and short-read genome sequencing technologies. American Journal of Human Genetics, 2021, 108, 919-928.	2.6	72
6	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4.	13.7	45
7	Evaluating drug targets through human loss-of-function genetic variation. Nature, 2020, 581, 459-464.	13.7	115
8	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	13.7	6,140
9	A structural variation reference for medical and population genetics. Nature, 2020, 581, 444-451.	13.7	614
10	Transcript expression-aware annotation improves rare variant interpretation. Nature, 2020, 581, 452-458.	13.7	142
11	Functional annotation of rare structural variation in the human brain. Nature Communications, 2020, 11, 2990.	5.8	32
12	Genome sequencing identifies a homozygous inversion disrupting QDPR as a cause for dihydropteridine reductase deficiency. Molecular Genetics & Genomic Medicine, 2020, 8, e1154.	0.6	8
13	Whole-Genome Sequencing to Characterize Monogenic and Polygenic Contributions in Patients Hospitalized With Early-Onset Myocardial Infarction. Circulation, 2019, 139, 1593-1602.	1.6	213
14	Primary cilia defects causing mitral valve prolapse. Science Translational Medicine, 2019, 11, .	5.8	76
15	Multi-platform discovery of haplotype-resolved structural variation in human genomes. Nature Communications, 2019, 10, 1784.	5.8	636
16	Next Generation Sequencing of Prenatal Structural Chromosomal Rearrangements Using Large-Insert Libraries. Methods in Molecular Biology, 2019, 1885, 251-265.	0.4	0
17	Dissecting the Causal Mechanism of X-Linked Dystonia-Parkinsonism by Integrating Genome and Transcriptome Assembly. Cell, 2018, 172, 897-909.e21.	13.5	163
18	An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. Nature Genetics, 2018, 50, 727-736.	9.4	235

#	Article	IF	Citations
19	Genome-wide de novo risk score implicates promoter variation in autism spectrum disorder. Science, 2018, 362, .	6.0	234
20	Risks and Recommendations in Prenatally Detected De Novo Balanced Chromosomal Rearrangements from Assessment of Long-Term Outcomes. American Journal of Human Genetics, 2018, 102, 1090-1103.	2.6	29
21	Cover Image, Volume 173A, Number 2, February 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	0.7	0
22	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
23	Defining the diverse spectrum of inversions, complex structural variation, and chromothripsis in the morbid humanÂgenome. Genome Biology, 2017, 18, 36.	3.8	159
24	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45.	9.4	251
25	Implication of LRRC4C and DPP6 in neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2017, 173, 395-406.	0.7	40
26	Indexcov: fast coverage quality control for whole-genome sequencing. GigaScience, 2017, 6, 1-6.	3.3	36
27	Estrogen-related receptor gamma implicated in a phenotype including hearing loss and mild developmental delay. European Journal of Human Genetics, 2016, 24, 1622-1626.	1.4	12
28	Structural Chromosomal Rearrangements Require Nucleotide-Level Resolution: Lessons from Next-Generation Sequencing in Prenatal Diagnosis. American Journal of Human Genetics, 2016, 99, 1015-1033.	2.6	53
29	Engineering microdeletions and microduplications by targeting segmental duplications with CRISPR. Nature Neuroscience, 2016, 19, 517-522.	7.1	72
30	Paired-Duplication Signatures Mark Cryptic Inversions and Other Complex Structural Variation. American Journal of Human Genetics, 2015, 97, 170-176.	2.6	45
31	Loss of Î´-catenin function in severe autism. Nature, 2015, 520, 51-56.	13.7	145
32	Cryptic and Complex Chromosomal Aberrations in Early-Onset Neuropsychiatric Disorders. American Journal of Human Genetics, 2014, 95, 454-461.	2.6	45
33	Multifactor dimensionality reduction reveals a three-locus epistatic interaction associated with susceptibility to pulmonary tuberculosis. BioData Mining, 2013, 6, 4.	2.2	34
34	An information-gain approach to detecting three-way epistatic interactions in genetic association studies. Journal of the American Medical Informatics Association: JAMIA, 2013, 20, 630-636.	2.2	69
35	Dissecting the Causal Mechanism of X-Linked Dystonia-Parkinsonism by Integrating Genome and Transcriptome Assembly. SSRN Electronic Journal, 0, , .	0.4	0