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List of Publications by Year in descending order

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Version: 2024-02-01

26
papers

5,565
citations

304368

22
h-index

552369

26
g-index

36
all docs

36
docs citations

36
times ranked

11072
citing authors

#	ARTICLE	IF	CITATIONS
1	Identification of Functional Elements and Regulatory Circuits by <i>Drosophila</i> modENCODE. <i>Science</i> , 2010, 330, 1787-1797.	6.0	1,124
2	Integrative Analysis of the <i>Caenorhabditis elegans</i> Genome by the modENCODE Project. <i>Science</i> , 2010, 330, 1775-1787.	6.0	912
3	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. <i>Human Mutation</i> , 2015, 36, 915-921.	1.1	390
4	Improved exome prioritization of disease genes through cross-species phenotype comparison. <i>Genome Research</i> , 2014, 24, 340-348.	2.4	300
5	Next-generation diagnostics and disease-gene discovery with the Exomiser. <i>Nature Protocols</i> , 2015, 10, 2004-2015.	5.5	296
6	Emergence and rapid transmission of SARS-CoV-2 B.1.1.7 in the United States. <i>Cell</i> , 2021, 184, 2587-2594.e7.	13.5	285
7	Linking Human Diseases to Animal Models Using Ontology-Based Phenotype Annotation. <i>PLoS Biology</i> , 2009, 7, e1000247.	2.6	247
8	A Whole-Genome Analysis Framework for Effective Identification of Pathogenic Regulatory Variants in Mendelian Disease. <i>American Journal of Human Genetics</i> , 2016, 99, 595-606.	2.6	223
9	The Human Phenotype Ontology: Semantic Unification of Common and Rare Disease. <i>American Journal of Human Genetics</i> , 2015, 97, 111-124.	2.6	203
10	Wastewater sequencing reveals early cryptic SARS-CoV-2 variant transmission. <i>Nature</i> , 2022, 609, 101-108.	13.7	200
11	Deletions of chromosomal regulatory boundaries are associated with congenital disease. <i>Genome Biology</i> , 2014, 15, 423.	3.8	144
12	FER-1 regulates Ca ²⁺ -mediated membrane fusion during <i>C. elegans</i> spermatogenesis. <i>Journal of Cell Science</i> , 2006, 119, 2552-2562.	1.2	126
13	modMine: flexible access to modENCODE data. <i>Nucleic Acids Research</i> , 2012, 40, D1082-D1088.	6.5	126
14	Population genetic screening efficiently identifies carriers of autosomal dominant diseases. <i>Nature Medicine</i> , 2020, 26, 1235-1239.	15.2	121
15	PhenomeCentral: A Portal for Phenotypic and Genotypic Matchmaking of Patients with Rare Genetic Diseases. <i>Human Mutation</i> , 2015, 36, 931-940.	1.1	107
16	Genome-wide rare variant analysis for thousands of phenotypes in over 70,000 exomes from two cohorts. <i>Nature Communications</i> , 2020, 11, 542.	5.8	101
17	Computational evaluation of exome sequence data using human and model organism phenotypes improves diagnostic efficiency. <i>Genetics in Medicine</i> , 2016, 18, 608-617.	1.1	85
18	Navigating the Phenotype Frontier: The Monarch Initiative. <i>Genetics</i> , 2016, 203, 1491-1495.	1.2	65

#	ARTICLE	IF	CITATIONS
19	SARS-CoV-2 variant Delta rapidly displaced variant Alpha in the United States and led to higher viral loads. <i>Cell Reports Medicine</i> , 2022, 3, 100564.	3.3	61
20	Pathogenic variants in actionable MODY genes are associated with type 2 diabetes. <i>Nature Metabolism</i> , 2020, 2, 1126-1134.	5.1	43
21	Use of Model Organism and Disease Databases to Support Matchmaking for Human Disease Gene Discovery. <i>Human Mutation</i> , 2015, 36, 979-984.	1.1	36
22	The modENCODE Data Coordination Center: lessons in harvesting comprehensive experimental details. <i>Database: the Journal of Biological Databases and Curation</i> , 2011, 2011, bar023.	1.4	32
23	Genetic counseling, 2030: An on-demand service tailored to the needs of a price conscious, genetically literate, and busy world. <i>Journal of Genetic Counseling</i> , 2019, 28, 456-465.	0.9	14
24	Positive predictive value highlights four novel candidates for actionable genetic screening from analysis of 220,000 clinicogenomic records. <i>Genetics in Medicine</i> , 2021, 23, 2300-2308.	1.1	13
25	Using symptom-based case predictions to identify host genetic factors that contribute to COVID-19 susceptibility. <i>PLoS ONE</i> , 2021, 16, e0255402.	1.1	6
26	Comprehensive Allele Genotyping in Critical Pharmacogenes Reduces Residual Clinical Risk in Diverse Populations. <i>Clinical Pharmacology and Therapeutics</i> , 2021, 110, 759-767.	2.3	4