

Max Schubach

List of Publications by Year in descending order

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Version: 2024-02-01

36
papers

2,499
citations

411340

20
h-index

466096

32
g-index

44
all docs

44
docs citations

44
times ranked

6031
citing authors

#	ARTICLE	IF	CITATIONS
1	CADD-Splice“improving genome-wide variant effect prediction using deep learning-derived splice scores. <i>Genome Medicine</i> , 2021, 13, 31.	3.6	375
2	Targeted next generation sequencing as a diagnostic tool in epileptic disorders. <i>Epilepsia</i> , 2012, 53, 1387-1398.	2.6	299
3	Next-generation diagnostics and disease-gene discovery with the Exomiser. <i>Nature Protocols</i> , 2015, 10, 2004-2015.	5.5	296
4	Panel-based next generation sequencing as a reliable and efficient technique to detect mutations in unselected patients with retinal dystrophies. <i>European Journal of Human Genetics</i> , 2014, 22, 99-104.	1.4	229
5	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
6	Saturation mutagenesis of twenty disease-associated regulatory elements at single base-pair resolution. <i>Nature Communications</i> , 2019, 10, 3583.	5.8	152
7	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. <i>Genome Biology</i> , 2014, 15, R53.	13.9	101
8	Mutation Detection in Patients with Retinal Dystrophies Using Targeted Next Generation Sequencing. <i>PLoS ONE</i> , 2016, 11, e0145951.	1.1	91
9	Further delineation of the SATB2 phenotype. <i>European Journal of Human Genetics</i> , 2014, 22, 1034-1039.	1.4	74
10	Characterization of glycosylphosphatidylinositol biosynthesis defects by clinical features, flow cytometry, and automated image analysis. <i>Genome Medicine</i> , 2018, 10, 3.	3.6	67
11	lentiMPRA and MPRAflow for high-throughput functional characterization of gene regulatory elements. <i>Nature Protocols</i> , 2020, 15, 2387-2412.	5.5	65
12	Imbalance-Aware Machine Learning for Predicting Rare and Common Disease-Associated Non-Coding Variants. <i>Scientific Reports</i> , 2017, 7, 2959.	1.6	63
13	PEDIA: prioritization of exome data by image analysis. <i>Genetics in Medicine</i> , 2019, 21, 2807-2814.	1.1	58
14	From ventriculomegaly to severe muscular atrophy: Expansion of the clinical spectrum related to mutations in AIFM1. <i>Mitochondrion</i> , 2015, 21, 12-18.	1.6	51
15	Integration of multiple epigenomic marks improves prediction of variant impact in saturation mutagenesis reporter assay. <i>Human Mutation</i> , 2019, 40, 1280-1291.	1.1	46
16	Haploinsufficiency of the Notch Ligand DLL1 Causes Variable Neurodevelopmental Disorders. <i>American Journal of Human Genetics</i> , 2019, 105, 631-639.	2.6	42
17	Loss-of-function variants in HIVEP2 are a cause of intellectual disability. <i>European Journal of Human Genetics</i> , 2016, 24, 556-561.	1.4	36
18	Whole exome sequencing of microdissected splenic marginal zone lymphoma: a study to discover novel tumor-specific mutations. <i>BMC Cancer</i> , 2015, 15, 773.	1.1	33

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19	Immune monitoring and TCR sequencing of CD4 T cells in a long term responsive patient with metastasized pancreatic ductal carcinoma treated with individualized, neoepitope-derived multipeptide vaccines: a case report. <i>Journal of Translational Medicine</i> , 2018, 16, 23.	1.8	30
20	StrÄ,mmme Syndrome Is a Ciliary Disorder Caused by Mutations in <i>CENPF</i> . <i>Human Mutation</i> , 2016, 37, 359-363.	1.1	27
21	Prediction of Human Phenotype Ontology terms by means of hierarchical ensemble methods. <i>BMC Bioinformatics</i> , 2017, 18, 449.	1.2	22
22	Multisite de novo mutations in human offspring after paternal exposure to ionizing radiation. <i>Scientific Reports</i> , 2018, 8, 14611.	1.6	22
23	Expanding the phenotype of a recurrent <i>de novo</i> variant in <i>PACS1</i> causing intellectual disability. <i>Clinical Genetics</i> , 2015, 88, 300-302.	1.0	17
24	Germline PTPN11 and somatic PIK3CA variant in a boy with megalencephaly-capillary malformation syndrome (MCAP) - pure coincidence?. <i>European Journal of Human Genetics</i> , 2015, 23, 409-412.	1.4	17
25	Alternate-locus aware variant calling in whole genome sequencing. <i>Genome Medicine</i> , 2016, 8, 130.	3.6	16
26	Short clones or long clones? A simulation study on the use of paired reads in metagenomics. <i>BMC Bioinformatics</i> , 2010, 11, S12.	1.2	11
27	parSMURF, a high-performance computing tool for the genome-wide detection of pathogenic variants. <i>GigaScience</i> , 2020, 9, .	3.3	11
28	The impact of different negative training data on regulatory sequence predictions. <i>PLoS ONE</i> , 2020, 15, e0237412.	1.1	4
29	Ensembling Descendant Term Classifiers to Improve Gene - Abnormal Phenotype Predictions. <i>Lecture Notes in Computer Science</i> , 2019, , 70-80.	1.0	2
30	Bayesian Optimization Improves Tissue-Specific Prediction of Active Regulatory Regions with Deep Neural Networks. <i>Lecture Notes in Computer Science</i> , 2020, , 600-612.	1.0	1
31	The impact of different negative training data on regulatory sequence predictions. , 2020, 15, e0237412.		0
32	The impact of different negative training data on regulatory sequence predictions. , 2020, 15, e0237412.		0
33	The impact of different negative training data on regulatory sequence predictions. , 2020, 15, e0237412.		0
34	The impact of different negative training data on regulatory sequence predictions. , 2020, 15, e0237412.		0
35	The impact of different negative training data on regulatory sequence predictions. , 2020, 15, e0237412.		0
36	The impact of different negative training data on regulatory sequence predictions. , 2020, 15, e0237412.		0