Max Schubach

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3368882/publications.pdf

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-Spliceâ€"improving genome-wide variant effect prediction using deep learning-derived splice scores. Genome Medicine, 2021, 13, 31.	3.6	375
2	parSMURF, a high-performance computing tool for the genome-wide detection of pathogenic variants. GigaScience, 2020, 9, .	3.3	11
3	lentiMPRA and MPRAflow for high-throughput functional characterization of gene regulatory elements. Nature Protocols, 2020, 15, 2387-2412.	5.5	65
4	Bayesian Optimization Improves Tissue-Specific Prediction of Active Regulatory Regions with Deep Neural Networks. Lecture Notes in Computer Science, 2020, , 600-612.	1.0	1
5	The impact of different negative training data on regulatory sequence predictions. PLoS ONE, 2020, 15, e0237412.	1.1	4
6	The impact of different negative training data on regulatory sequence predictions. , 2020, 15, e0237412.		0
7	The impact of different negative training data on regulatory sequence predictions. , 2020, 15, e0237412.		0
8	The impact of different negative training data on regulatory sequence predictions. , 2020, 15, e0237412.		0
9	The impact of different negative training data on regulatory sequence predictions. , 2020, 15, e0237412.		0
10	The impact of different negative training data on regulatory sequence predictions. , 2020, 15, e0237412.		0
11	The impact of different negative training data on regulatory sequence predictions. , 2020, 15, e0237412.		0
12	Saturation mutagenesis of twenty disease-associated regulatory elements at single base-pair resolution. Nature Communications, 2019, 10, 3583.	5.8	152
13	Haploinsufficiency of the Notch Ligand DLL1 Causes Variable Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 105, 631-639.	2.6	42
14	PEDIA: prioritization of exome data by image analysis. Genetics in Medicine, 2019, 21, 2807-2814.	1.1	58
15	Integration of multiple epigenomic marks improves prediction of variant impact in saturation mutagenesis reporter assay. Human Mutation, 2019, 40, 1280-1291.	1.1	46
16	Ensembling Descendant Term Classifiers to Improve Gene - Abnormal Phenotype Predictions. Lecture Notes in Computer Science, 2019, , 70-80.	1.0	2
17	Multisite de novo mutations in human offspring after paternal exposure to ionizing radiation. Scientific Reports, 2018, 8, 14611.	1.6	22
18	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. Journal of Translational Medicine, 2018, 16, 23.	1.8	30

#	Article	IF	Citations
19	Characterization of glycosylphosphatidylinositol biosynthesis defects by clinical features, flow cytometry, and automated image analysis. Genome Medicine, 2018, 10, 3.	3.6	67
20	Imbalance-Aware Machine Learning for Predicting Rare and Common Disease-Associated Non-Coding Variants. Scientific Reports, 2017, 7, 2959.	1.6	63
21	Prediction of Human Phenotype Ontology terms by means of hierarchical ensemble methods. BMC Bioinformatics, 2017, 18, 449.	1.2	22
22	Mutation Detection in Patients with Retinal Dystrophies Using Targeted Next Generation Sequencing. PLoS ONE, 2016, 11, e0145951.	1.1	91
23	Alternate-locus aware variant calling in whole genome sequencing. Genome Medicine, 2016, 8, 130.	3.6	16
24	StrÃ¸mme Syndrome Is a Ciliary Disorder Caused by Mutations inCENPF. Human Mutation, 2016, 37, 359-363.	1.1	27
25	A Whole-Genome Analysis Framework for Effective Identification of Pathogenic Regulatory Variants in Mendelian Disease. American Journal of Human Genetics, 2016, 99, 595-606.	2.6	223
26	Loss-of-function variants in HIVEP2 are a cause of intellectual disability. European Journal of Human Genetics, 2016, 24, 556-561.	1.4	36
27	Expanding the phenotype of a recurrent de novo variant in <scp>PACS1</scp> causing intellectual disability. Clinical Genetics, 2015, 88, 300-302.	1.0	17
28	Whole exome sequencing of microdissected splenic marginal zone lymphoma: a study to discover novel tumor-specific mutations. BMC Cancer, 2015, 15, 773.	1.1	33
29	Next-generation diagnostics and disease-gene discovery with the Exomiser. Nature Protocols, 2015, 10, 2004-2015.	5.5	296
30	From ventriculomegaly to severe muscular atrophy: Expansion of the clinical spectrum related to mutations in AIFM1. Mitochondrion, 2015, 21, 12-18.	1.6	51
31	Germline PTPN11 and somatic PIK3CA variant in a boy with megalencephaly-capillary malformation syndrome (MCAP) - pure coincidence?. European Journal of Human Genetics, 2015, 23, 409-412.	1.4	17
32	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. Genome Biology, 2014, 15, R53.	13.9	101
33	Further delineation of the SATB2 phenotype. European Journal of Human Genetics, 2014, 22, 1034-1039.	1.4	74
34	Panel-based next generation sequencing as a reliable and efficient technique to detect mutations in unselected patients with retinal dystrophies. European Journal of Human Genetics, 2014, 22, 99-104.	1.4	229
35	Targeted next generation sequencing as a diagnostic tool in epileptic disorders. Epilepsia, 2012, 53, 1387-1398.	2.6	299
36	Short clones or long clones? A simulation study on the use of paired reads in metagenomics. BMC Bioinformatics, 2010, 11, S12.	1.2	11