Max Schubach

List of Publications by Year
in descending order

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36	2,499	361413	414414
papers	citations	20 h-index	32 g-index

44	44	44	5559
all docs	docs citations	times ranked	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.	8.2	375
2	Targeted next generation sequencing as a diagnostic tool in epileptic disorders. Epilepsia, 2012, 53, 1387-1398.	5.1	299
3	Next-generation diagnostics and disease-gene discovery with the Exomiser. Nature Protocols, 2015, 10, 2004-2015.	12.0	296
4	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.	2.8	229
5	A Whole-Genome Analysis Framework for Effective Identification of Pathogenic Regulatory Variants in Mendelian Disease. American Journal of Human Genetics, 2016, 99, 595-606.	6.2	223
6	Saturation mutagenesis of twenty disease-associated regulatory elements at single base-pair resolution. Nature Communications, 2019, 10, 3583.	12.8	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. Genome Biology, 2014, 15, R53.	9.6	101
8	Mutation Detection in Patients with Retinal Dystrophies Using Targeted Next Generation Sequencing. PLoS ONE, 2016, 11, e0145951.	2.5	91
9	Further delineation of the SATB2 phenotype. European Journal of Human Genetics, 2014, 22, 1034-1039.	2.8	74
10	Characterization of glycosylphosphatidylinositol biosynthesis defects by clinical features, flow cytometry, and automated image analysis. Genome Medicine, 2018, 10, 3.	8.2	67
11	lentiMPRA and MPRAflow for high-throughput functional characterization of gene regulatory elements. Nature Protocols, 2020, 15, 2387-2412.	12.0	65
12	Imbalance-Aware Machine Learning for Predicting Rare and Common Disease-Associated Non-Coding Variants. Scientific Reports, 2017, 7, 2959.	3.3	63
13	PEDIA: prioritization of exome data by image analysis. Genetics in Medicine, 2019, 21, 2807-2814.	2.4	58
14	From ventriculomegaly to severe muscular atrophy: Expansion of the clinical spectrum related to mutations in AIFM1. Mitochondrion, 2015, 21, 12-18.	3.4	51
15	Integration of multiple epigenomic marks improves prediction of variant impact in saturation mutagenesis reporter assay. Human Mutation, 2019, 40, 1280-1291.	2.5	46
16	Haploinsufficiency of the Notch Ligand DLL1 Causes Variable Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 105, 631-639.	6.2	42
17	Loss-of-function variants in HIVEP2 are a cause of intellectual disability. European Journal of Human Genetics, 2016, 24, 556-561.	2.8	36
18	Whole exome sequencing of microdissected splenic marginal zone lymphoma: a study to discover novel tumor-specific mutations. BMC Cancer, 2015, 15, 773.	2.6	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. Journal of Translational Medicine, 2018, 16, 23.	4.4	30
20	StrÃ¸mme Syndrome Is a Ciliary Disorder Caused by Mutations inCENPF. Human Mutation, 2016, 37, 359-363.	2.5	27
21	Prediction of Human Phenotype Ontology terms by means of hierarchical ensemble methods. BMC Bioinformatics, 2017, 18, 449.	2.6	22
22	Multisite de novo mutations in human offspring after paternal exposure to ionizing radiation. Scientific Reports, 2018, 8, 14611.	3.3	22
23	Expanding the phenotype of a recurrent de novo variant in <scp>PACS1</scp> causing intellectual disability. Clinical Genetics, 2015, 88, 300-302.	2.0	17
24	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.	2.8	17
25	Alternate-locus aware variant calling in whole genome sequencing. Genome Medicine, 2016, 8, 130.	8.2	16
26	Short clones or long clones? A simulation study on the use of paired reads in metagenomics. BMC Bioinformatics, 2010, 11, S12.	2.6	11
27	parSMURF, a high-performance computing tool for the genome-wide detection of pathogenic variants. GigaScience, 2020, 9, .	6.4	11
28	The impact of different negative training data on regulatory sequence predictions. PLoS ONE, 2020, 15, e0237412.	2.5	4
29	Ensembling Descendant Term Classifiers to Improve Gene - Abnormal Phenotype Predictions. Lecture Notes in Computer Science, 2019, , 70-80.	1.3	2
30	Bayesian Optimization Improves Tissue-Specific Prediction of Active Regulatory Regions with Deep Neural Networks. Lecture Notes in Computer Science, 2020, , 600-612.	1.3	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
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36	The impact of different negative training data on regulatory sequence predictions. , 2020, 15, e0237412.		0