Mahdi Sarmady

List of Publications by Year
in descending order

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30 papers	1,269 citations	516710 16 h-index	454955 30 g-index
39 all docs	39 docs citations	39 times ranked	3490 citing authors

#	Article	IF	Citations
1	Molecular Diagnostic Outcomes from 700 Cases. Journal of Molecular Diagnostics, 2022, 24, 274-286.	2.8	7
2	Evaluating the impact of in silico predictors on clinical variant classification. Genetics in Medicine, 2022, 24, 924-930.	2.4	20
3	Using Machine Learning to Identify True Somatic Variants from Next-Generation Sequencing. Clinical Chemistry, 2020, 66, 239-246.	3.2	7
4	Diagnosing Cornelia de Lange syndrome and related neurodevelopmental disorders using RNA sequencing. Genetics in Medicine, 2020, 22, 927-936.	2.4	34
5	AnthOligo: automating the design of oligonucleotides for capture/enrichment technologies. Bioinformatics, 2020, 36, 4353-4356.	4.1	4
6	Phen2Gene: rapid phenotype-driven gene prioritization for rare diseases. NAR Genomics and Bioinformatics, 2020, 2, lqaa032.	3.2	45
7	Utilizing nanopore sequencing technology for the rapid and comprehensive characterization of eleven HLA loci; addressing the need for deceased donor expedited HLA typing. Human Immunology, 2020, 81, 413-422.	2.4	37
8	Genetic variant pathogenicity prediction trained using disease-specific clinical sequencing data sets. Genome Research, 2019, 29, 1144-1151.	5.5	19
9	Clinical utility of custom-designed NGS panel testing in pediatric tumors. Genome Medicine, 2019, 11, 32.	8.2	79
10	Use of a Dynamic Genetic Testing Approach for Childhood-Onset Epilepsy. JAMA Network Open, 2019, 2, e192129.	5.9	45
11	A comparison of survival analysis methods for cancer gene expression RNA-Sequencing data. Cancer Genetics, 2019, 235-236, 1-12.	0.4	11
12	A mutation update for the PCDH19 gene causing early-onset epilepsy in females with an unusual expression pattern. Human Mutation, 2019, 40, 243-257.	2.5	23
13	Automated Clinical Exome Reanalysis Reveals Novel Diagnoses. Journal of Molecular Diagnostics, 2019, 21, 38-48.	2.8	68
14	Rapid and accurate interpretation of clinical exomes using Phenoxome: a computational phenotype-driven approach. European Journal of Human Genetics, 2019, 27, 612-620.	2.8	17
15	AUDIOME: a tiered exome sequencingâ€“based comprehensive gene panel for the diagnosis of heterogeneous nonsyndromic sensorineural hearing loss. Genetics in Medicine, 2018, 20, 1600-1608.	2.4	27
16	Novel findings with reassessment of exome data: implications for validation testing and interpretation of genomic data. Genetics in Medicine, 2018, 20, 329-336.	2.4	28
17	Characterizing reduced coverage regions through comparison of exome and genome sequencing data across 10 centers. Genetics in Medicine, 2018, 20, 855-866.	2.4	22
18	Need for Automated Interactive Genomic Interpretation and Ongoing Reanalysis. JAMA Pediatrics, 2018, 172, 1113.	6.2	12

#	Article	IF	Citations
19	The Development and Validation of Clinical Exome-Based Panels Using ExomeSlicer. Journal of Molecular Diagnostics, 2018, 20, 643-652.	2.8	16
20	Utility and limitations of exome sequencing as a genetic diagnostic tool for children with hearing loss. Genetics in Medicine, 2018, 20, 1663-1676.	2.4	26
21	Transcriptome analysis of IL-10-stimulated (M2c) macrophages by next-generation sequencing. Immunobiology, 2017, 222, 847-856.	1.9	142
22	Using large sequencing data sets to refine intragenic disease regions and prioritize clinical variant interpretation. Genetics in Medicine, 2017, 19, 496-504.	2.4	15
23	Utility and limitations of exome sequencing as a genetic diagnostic tool for conditions associated with pediatric sudden cardiac arrest/sudden cardiac death. Human Genomics, 2015, 9, 15.	2.9	5
24	Exome sequencing expands the mechanism of SOX5â€associated intellectual disability: A case presentation with review of soxâ€related disorders. American Journal of Medical Genetics, Part A, 2015, 167, 2548-2554.	1.2	26
25	Exome Sequencing Analysis Reveals Variants in Primary Immunodeficiency Genes in Patients With Very Early Onset Inflammatory Bowel Disease. Gastroenterology, 2015, 149, 1415-1424.	1.3	99
26	mtDNA Variation and Analysis Using Mitomap and Mitomaster. Current Protocols in Bioinformatics, 2013, 44, 1.23.1-26.	25.8	390
27	Efficient digest of high-throughput sequencing data in a reproducible report. BMC Bioinformatics, 2013, 14, S3.	2.6	6
28	Bamchop: A bioinformatics utility to summarize and visualize exome and other types of targeted resequencing data. , 2012, , .		0
29	HIV Protein Sequence Hotspots for Crosstalk with Host Hub Proteins. PLoS ONE, 2011, 6, e23293.	2.5	23
30	Sequence- and Interactome-Based Prediction of Viral Protein Hotspots Targeting Host Proteins: A Case Study for HIV Nef. PLoS ONE, 2011, 6, e20735.	2.5	11