Jana Marie Schwarz

List of Publications by Year
in descending order

15 papers	6,183 citations	1170033 9 h-index	1113639 15 g-index
17 all docs	17 docs citations	17 times ranked	18418 citing authors

#	Article	IF	Citations
1	MutationTaster2021. Nucleic Acids Research, 2021, 49, W446-W451.	6.5	122
2	Public data sources for regulatory genomic features. Medizinische Genetik, 2021, 33, 167-177.	0.1	1
3	Novel sequencing technologies and bioinformatic tools for deciphering the non-coding genome. Medizinische Genetik, 2021, 33, 133-145.	0.1	1
4	A new homozygous HERC1 gain-of-function variant in MDFPMR syndrome leads to mTORC1 hyperactivation and reduced autophagy during cell catabolism. Molecular Genetics and Metabolism, 2020, 131, 126-134.	0.5	6
5	A spontaneous missense mutation in the chromodomain helicase DNAâ€binding protein 8 (CHD8) gene: a novel association with congenital myasthenic syndrome. Neuropathology and Applied Neurobiology, 2020, 46, 588-601.	1.8	6
6	RegulationSpotter: annotation and interpretation of extratranscriptic DNA variants. Nucleic Acids Research, 2019, 47, W106-W113.	6.5	17
7	MutationDistiller: user-driven identification of pathogenic DNA variants. Nucleic Acids Research, 2019, 47, W114-W120.	6.5	37
8	Phenotero: Annotate as you write. Clinical Genetics, 2019, 95, 287-292.	1.0	3
9	Recessive mutation in EXOSC3 associates with mitochondrial dysfunction and pontocerebellar hypoplasia. Mitochondrion, 2017, 37, 46-54.	1.6	26
10	A systematic, large-scale comparison of transcription factor binding site models. BMC Genomics, 2016, 17, 388.	1.2	15
11	MutationTaster2: mutation prediction for the deep-sequencing age. Nature Methods, 2014, 11, 361-362.	9.0	3,203
12	CNVinspector:a web-based tool for the interactive evaluation of copy number variations in single patients and in cohorts. Journal of Medical Genetics, 2013, 50, 529-533.	1.5	3
13	Systematic Comparison of Three Methods for Fragmentation of Long-Range PCR Products for Next Generation Sequencing. PLoS ONE, 2011, 6, e28240.	1.1	106
14	MutationTaster evaluates disease-causing potential of sequence alterations. Nature Methods, 2010, 7, 575-576.	9.0	2,538
15	GeneDistillerâ€”Distilling Candidate Genes from Linkage Intervals. PLoS ONE, 2008, 3, e3874.	1.1	98