## Joke Reumers

List of Publications by Year in descending order

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LOKE RELIMEDS

#	Article	lF	CITATIONS
1	Genetically Engineered iPSC-Derived FTDP-17 MAPT Neurons Display Mutation-Specific Neurodegenerative and Neurodevelopmental Phenotypes. Stem Cell Reports, 2018, 11, 363-379.	2.3	43
2	A random effects model for the identification of differential splicing (REIDS) using exon and HTA arrays. BMC Bioinformatics, 2017, 18, 273.	1.2	2
3	Identification of cis-regulatory mutations generating de novo edges in personalized cancer gene regulatory networks. Genome Medicine, 2017, 9, 80.	3.6	17
4	Halvade-RNA: Parallel variant calling from transcriptomic data using MapReduce. PLoS ONE, 2017, 12, e0174575.	1.1	17
5	Performance Analysis of a Parallel, Multi-node Pipeline for DNA Sequencing. Lecture Notes in Computer Science, 2016, , 233-242.	1.0	2
6	elPrep: High-Performance Preparation of Sequence Alignment/Map Files for Variant Calling. PLoS ONE, 2015, 10, e0132868.	1.1	22
7	Halvade: scalable sequence analysis with MapReduce. Bioinformatics, 2015, 31, 2482-2488.	1.8	69
8	QQ-SNV: single nucleotide variant detection at low frequency by comparing the quality quantiles. BMC Bioinformatics, 2015, 16, 379.	1.2	3
9	ViVaMBC: estimating viral sequence variation in complex populations from illumina deep-sequencing data using model-based clustering. BMC Bioinformatics, 2015, 16, 59.	1.2	12
10	Performance assessment of the Illumina massively parallel sequencing platform for deep sequencing analysis of viral minority variants. Journal of Virological Methods, 2015, 221, 29-38.	1.0	26
11	VirVarSeq: a low-frequency virus variant detection pipeline for Illumina sequencing using adaptive base-calling accuracy filtering. Bioinformatics, 2015, 31, 94-101.	1.8	47
12	Genome dynamics of the human embryonic kidney 293 lineage in response to cell biology manipulations. Nature Communications, 2014, 5, 4767.	5.8	421
13	Mismatch repair deficiency endows tumors with a unique mutation signature and sensitivity to DNA double-strand breaks. ELife, 2014, 3, e02725.	2.8	71
14	Efficiency of whole genome amplification of single circulating tumor cells enriched by CellSearch and sorted by FACS. Genome Medicine, 2013, 5, 106.	3.6	79
15	Pipit: visualizing functional impacts of structural variations. Bioinformatics, 2013, 29, 2206-2207.	1.8	1
16	Whole-genome sequencing reveals a coding non-pathogenic variant tagging a non-coding pathogenic hexanucleotide repeat expansion in C9orf72 as cause of amyotrophic lateral sclerosis. Human Molecular Genetics, 2012, 21, 2412-2419.	1.4	33
17	Optimized filtering reduces the error rate in detecting genomic variants by short-read sequencing. Nature Biotechnology, 2012, 30, 61-68.	9.4	211
18	VEGF pathway genetic variants as biomarkers of treatment outcome with bevacizumab: an analysis of data from the AVITA and AVOREN randomised trials. Lancet Oncology, The, 2012, 13, 724-733.	5.1	174

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#	Article	IF	CITATIONS
19	SNPeffect 4.0: on-line prediction of molecular and structural effects of protein-coding variants. Nucleic Acids Research, 2012, 40, D935-D939.	6.5	235
20	α-Galactosidase Aggregation Is a Determinant of Pharmacological Chaperone Efficacy on Fabry Disease Mutants. Journal of Biological Chemistry, 2012, 287, 28386-28397.	1.6	31
21	Gain of function of mutant p53 by coaggregation with multiple tumor suppressors. Nature Chemical Biology, 2011, 7, 285-295.	3.9	450
22	An Evolutionary Trade-Off between Protein Turnover Rate and Protein Aggregation Favors a Higher Aggregation Propensity in Fast Degrading Proteins. PLoS Computational Biology, 2011, 7, e1002090.	1.5	50
23	Sequencing of DISC1 Pathway Genes Reveals Increased Burden of Rare Missense Variants in Schizophrenia Patients from a Northern Swedish Population. PLoS ONE, 2011, 6, e23450.	1.1	46
24	Assessing interactions between the associations of common genetic susceptibility variants, reproductive history and body mass index with breast cancer risk in the breast cancer association consortium: a combined case-control study. Breast Cancer Research, 2010, 12, R110.	2.2	82
25	Exploring the sequence determinants of amyloid structure using position-specific scoring matrices. Nature Methods, 2010, 7, 237-242.	9.0	566
26	PepX: a structural database of non-redundant protein–peptide complexes. Nucleic Acids Research, 2010, 38, D545-D551.	6.5	102
27	Multiple Evolutionary Mechanisms Reduce Protein Aggregation~!2009-04-21~!2009-07-09~!2010-01-02~!. The Open Biology Journal, 2010, 2, 176-184.	0.5	7
28	Using structural bioinformatics to investigate the impact of non synonymous SNPs and disease mutations: scope and limitations. BMC Bioinformatics, 2009, 10, S9.	1.2	22
29	Protein sequences encode safeguards against aggregation. Human Mutation, 2009, 30, 431-437.	1.1	86
30	Genetic variability in the mitochondrial serine protease <i>HTRA2</i> contributes to risk for Parkinson disease. Human Mutation, 2008, 29, 832-840.	1.1	107
31	Joint annotation of coding and non-coding single nucleotide polymorphisms and mutations in the SNPeffect and PupaSuite databases. Nucleic Acids Research, 2008, 36, D825-D829.	6.5	60
32	Reconstruction of Protein Backbones from the BriX Collection of Canonical Protein Fragments. PLoS Computational Biology, 2008, 4, e1000083.	1.5	42
33	SNPeffect v2.0: a new step in investigating the molecular phenotypic effects of human non-synonymous SNPs. Bioinformatics, 2006, 22, 2183-2185.	1.8	80
34	PupaSuite: finding functional single nucleotide polymorphisms for large-scale genotyping purposes. Nucleic Acids Research, 2006, 34, W621-W625.	6.5	194
35	SNPeffect: a database mapping molecular phenotypic effects of human non-synonymous coding SNPs. Nucleic Acids Research, 2004, 33, D527-D532.	6.5	136