Robin van der Lee

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/1145072/publications.pdf

Version: 2024-02-01

28 papers 5,918 citations

16 h-index 27 g-index

32 all docs 32 docs citations

times ranked

32

15253 citing authors

#	Article	IF	CITATIONS
1	RevUP: an online scoring system for regulatory variants implicated in rare diseases. Bioinformatics, 2022, 38, 2664-2666.	4.1	O
2	LY-CoV1404 (bebtelovimab) potently neutralizes SARS-CoV-2 variants. Cell Reports, 2022, 39, 110812.	6.4	287
3	The role of clinical response to treatment in determining pathogenicity of genomic variants. Genetics in Medicine, 2021, 23, 581-585.	2.4	18
4	<i>De novo</i> stop-loss variants in <i>CLDN11</i> cause hypomyelinating leukodystrophy. Brain, 2021, 144, 411-419.	7.6	12
5	Response to Biesecker et al Genetics in Medicine, 2021, 23, 793-794.	2.4	O
6	Adult GAMT deficiency: A literature review and report of two siblings. Molecular Genetics and Metabolism Reports, 2021, 27, 100761.	1.1	6
7	JASPAR 2020: update of the open-access database of transcription factor binding profiles. Nucleic Acids Research, 2020, 48, D87-D92.	14.5	1,039
8	Association between fetal sex, birthweight percentile and adverse pregnancy outcome. Acta Obstetricia Et Gynecologica Scandinavica, 2020, 99, 48-58.	2.8	21
9	Ductus arteriosus and failed medical therapy. Journal of Neonatal-Perinatal Medicine, 2020, 13, 39-45.	0.8	1
10	Deregulated Regulators: Disease-Causing cis Variants in Transcription Factor Genes. Trends in Genetics, 2020, 36, 523-539.	6.7	26
11	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. American Journal of Human Genetics, 2019, 105, 534-548.	6.2	46
12	A Combined Mass Spectrometry and Data Integration Approach to Predict the Mitochondrial Poly(A) RNA Interacting Proteome. Frontiers in Cell and Developmental Biology, 2019, 7, 283.	3.7	2
13	CiliaCarta: An integrated and validated compendium of ciliary genes. PLoS ONE, 2019, 14, e0216705.	2.5	104
14	Understanding the pathobiology in patent ductus arteriosus in prematurityâ€"beyond prostaglandins and oxygen. Pediatric Research, 2019, 86, 28-38.	2.3	28
15	Strabismus in Children With Intellectual Disability: Part of a Broader Motor Control Phenotype?. Pediatric Neurology, 2019, 100, 87-91.	2.1	4
16	Curation and bioinformatic analysis of strabismus genes supports functional heterogeneity and proposes candidate genes with connections to RASopathies. Gene, 2019, 697, 213-226.	2.2	5
17	JASPAR 2018: update of the open-access database of transcription factor binding profiles and its web framework. Nucleic Acids Research, 2018, 46, D260-D266.	14.5	1,232
18	Probabilistic data integration identifies reliable gametocyte-specific proteins and transcripts in malaria parasites. Scientific Reports, 2018, 8, 410.	3.3	39

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19	The role of NeonatologistÂPerformed Echocardiography in the assessment and management of neonatal shock. Pediatric Research, 2018, 84, 57-67.	2.3	67
20	Genome-scale detection of positive selection in nine primates predicts human-virus evolutionary conflicts. Nucleic Acids Research, 2017, 45, 10634-10648.	14.5	76
21	Immunologic defects in severe mucocutaneous HSV-2 infections: Response to IFN-γ therapy. Journal of Allergy and Clinical Immunology, 2016, 138, 895-898.	2.9	6
22	TMEM107 recruits ciliopathy proteins to subdomains of the ciliary transition zone and causes JoubertÂsyndrome. Nature Cell Biology, 2016, 18, 122-131.	10.3	118
23	Transcriptome analysis of complex I-deficient patients reveals distinct expression programs for subunits and assembly factors of the oxidative phosphorylation system. BMC Genomics, 2015, 16, 691.	2.8	12
24	The RIG-I-like helicase receptor MDA5 (IFIH1) is involved in the host defense against Candida infections. European Journal of Clinical Microbiology and Infectious Diseases, 2015, 34, 963-974.	2.9	69
25	Integrative Genomics-Based Discovery of Novel Regulators of the Innate Antiviral Response. PLoS Computational Biology, 2015, 11, e1004553.	3.2	25
26	Intrinsically Disordered Segments Affect Protein Half-Life in the Cell and during Evolution. Cell Reports, 2014, 8, 1832-1844.	6.4	192
27	Classification of Intrinsically Disordered Regions and Proteins. Chemical Reviews, 2014, 114, 6589-6631.	47.7	1,618
28	Intrinsically disordered proteins: regulation and disease. Current Opinion in Structural Biology, 2011, 21, 432-440.	5.7	518