

Bernard J Pope

List of Publications by Year in descending order

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Version: 2024-02-01

54
papers

2,218
citations

471061

17
h-index

253896

43
g-index

62
all docs

62
docs citations

62
times ranked

4831
citing authors

#	ARTICLE	IF	CITATIONS
1	Long-read assembly and comparative evidence-based reanalysis of <i>Cryptosporidium</i> genome sequences reveal expanded transporter repertoire and duplication of entire chromosome ends including subtelomeric regions. <i>Genome Research</i> , 2022, 32, 203-213.	2.4	26
2	Phase 2 Study of Neoadjuvant FGFR Inhibition and Androgen Deprivation Therapy Prior to Prostatectomy. <i>Clinical Genitourinary Cancer</i> , 2022, 20, 452-458.	0.9	5
3	Identifying colorectal cancer caused by biallelic MUTYH pathogenic variants using tumor mutational signatures. <i>Nature Communications</i> , 2022, 13, .	5.8	15
4	Evaluating the utility of tumour mutational signatures for identifying hereditary colorectal cancer and polyposis syndrome carriers. <i>Gut</i> , 2021, 70, 2138-2149.	6.1	27
5	Germline and Tumor Sequencing as a Diagnostic Tool To Resolve Suspected Lynch Syndrome. <i>Journal of Molecular Diagnostics</i> , 2021, 23, 358-371.	1.2	12
6	MSH2-deficient prostate tumours have a distinct immune response and clinical outcome compared to MSH2-deficient colorectal or endometrial cancer. <i>Prostate Cancer and Prostatic Diseases</i> , 2021, 24, 1167-1180.	2.0	4
7	Rare germline variants in the AXIN2 gene in families with colonic polyposis and colorectal cancer. <i>Familial Cancer</i> , 2021, , 1.	0.9	5
8	Population-based estimates of breast cancer risk for carriers of pathogenic variants identified by gene-panel testing. <i>Npj Breast Cancer</i> , 2021, 7, 153.	2.3	10
9	SNPPar: identifying convergent evolution and other homoplasies from microbial whole-genome alignments. <i>Microbial Genomics</i> , 2021, 7, .	1.0	5
10	HiTIME: An efficient model-selection approach for the detection of unknown drug metabolites in LC-MS data. <i>SoftwareX</i> , 2020, 12, 100559.	1.2	3
11	Monoallelic NTHL1 Loss-of-Function Variants and Risk of Polyposis and Colorectal Cancer. <i>Gastroenterology</i> , 2020, 159, 2241-2243.e6.	0.6	20
12	Genetic testing in Poland and Ukraine: should comprehensive germline testing of <i>BRCA1</i> and <i>BRCA2</i> be recommended for women with breast and ovarian cancer?. <i>Genetical Research</i> , 2020, 102, e6.	0.3	12
13	Detection of ctDNA in plasma of patients with clinically localised prostate cancer is associated with rapid disease progression. <i>Genome Medicine</i> , 2020, 12, 72.	3.6	35
14	Mismatch repair gene pathogenic germline variants in a population-based cohort of breast cancer. <i>Familial Cancer</i> , 2020, 19, 197-202.	0.9	6
15	Rare germline genetic variants and risk of aggressive prostate cancer. <i>International Journal of Cancer</i> , 2020, 147, 2142-2149.	2.3	12
16	Hi-Plex2: a simple and robust approach to targeted sequencing-based genetic screening. <i>BioTechniques</i> , 2019, 67, 118-122.	0.8	11
17	Bionitio: demonstrating and facilitating best practices for bioinformatics command-line software. <i>GigaScience</i> , 2019, 8, .	3.3	13
18	Tumor mutational signatures in sebaceous skin lesions from individuals with Lynch syndrome. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e00781.	0.6	8

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19	Annotation of the <i>Giardia</i> proteome through structure-based homology and machine learning. <i>GigaScience</i> , 2019, 8, .	3.3	21
20	Is RNASEL:p.Glu265* a modifier of early-onset breast cancer risk for carriers of high-risk mutations?. <i>BMC Cancer</i> , 2018, 18, 165.	1.1	6
21	Somatic mutations of the coding microsatellites within the beta-2-microglobulin gene in mismatch repair-deficient colorectal cancers and adenomas. <i>Familial Cancer</i> , 2018, 17, 91-100.	0.9	21
22	Targeted massively parallel sequencing characterises the mutation spectrum of PALB2 in breast and ovarian cancer cases from Poland and Ukraine. <i>Familial Cancer</i> , 2018, 17, 345-349.	0.9	7
23	Hi-Plex for Simple, Accurate, and Cost-Effective Amplicon-based Targeted DNA Sequencing. <i>Methods in Molecular Biology</i> , 2018, 1712, 53-70.	0.4	2
24	Risk of colorectal cancer for carriers of a germ-line mutation in POLE or POLD1. <i>Genetics in Medicine</i> , 2018, 20, 890-895.	1.1	49
25	sEst: Accurate Sex-Estimation and Abnormality Detection in Methylation Microarray Data. <i>International Journal of Molecular Sciences</i> , 2018, 19, 3172.	1.8	8
26	FANCM and RECQL genetic variants and breast cancer susceptibility: relevance to South Poland and West Ukraine. <i>BMC Medical Genetics</i> , 2018, 19, 12.	2.1	20
27	Mutation screening of ACKR3 and COPS8 in kidney cancer cases from the CONFIRM study. <i>Familial Cancer</i> , 2017, 16, 411-416.	0.9	6
28	Single nucleotide-level mapping of DNA double-strand breaks in human HEK293T cells. <i>Genomics Data</i> , 2017, 11, 43-45.	1.3	0
29	Variant effect prediction tools assessed using independent, functional assay-based datasets: implications for discovery and diagnostics. <i>Human Genomics</i> , 2017, 11, 10.	1.4	68
30	A novel <i>Drosophila</i> injury model reveals severed axons are cleared through a Draper/MMP-1 signaling cascade. <i>ELife</i> , 2017, 6, .	2.8	47
31	Four simple recommendations to encourage best practices in research software. <i>F1000Research</i> , 2017, 6, 876.	0.8	88
32	Best practice data life cycle approaches for the life sciences. <i>F1000Research</i> , 2017, 6, 1618.	0.8	21
33	Best practice data life cycle approaches for the life sciences. <i>F1000Research</i> , 2017, 6, 1618.	0.8	23
34	Fine resolution mapping of double-strand break sites for human ribosomal DNA units. <i>Genomics Data</i> , 2016, 10, 19-21.	1.3	1
35	UNDR ROVER - a fast and accurate variant caller for targeted DNA sequencing. <i>BMC Bioinformatics</i> , 2016, 17, 165.	1.2	3
36	MethPat: a tool for the analysis and visualisation of complex methylation patterns obtained by massively parallel sequencing. <i>BMC Bioinformatics</i> , 2016, 17, 98.	1.2	22

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37	Exemplary multiplex bisulfite amplicon data used to demonstrate the utility of Methpat. <i>GigaScience</i> , 2015, 4, 55.	3.3	3
38	Abridged adapter primers increase the target scope of Hi-Plex. <i>BioTechniques</i> , 2015, 58, 33-6.	0.8	7
39	Mutation screening of PALB2 in clinically ascertained families from the Breast Cancer Family Registry. <i>Breast Cancer Research and Treatment</i> , 2015, 149, 547-554.	1.1	23
40	High-Resolution Twin-Ion Metabolite Extraction (HiTIME) Mass Spectrometry: Nontargeted Detection of Unknown Drug Metabolites by Isotope Labeling, Liquid Chromatography Mass Spectrometry, and Automated High-Performance Computing. <i>Analytical Chemistry</i> , 2015, 87, 4104-4109.	3.2	23
41	SRST2: Rapid genomic surveillance for public health and hospital microbiology labs. <i>Genome Medicine</i> , 2014, 6, 90.	3.6	953
42	ROVER variant caller: read-pair overlap considerate variant-calling software applied to PCR-based massively parallel sequencing datasets. <i>Source Code for Biology and Medicine</i> , 2014, 9, 3.	1.7	12
43	Rare Mutations in <i>RINT1</i> Predispose Carriers to Breast and Lynch Syndrome "Spectrum Cancers. <i>Cancer Discovery</i> , 2014, 4, 804-815.	7.7	44
44	FAVR (Filtering and Annotation of Variants that are Rare): methods to facilitate the analysis of rare germline genetic variants from massively parallel sequencing datasets. <i>BMC Bioinformatics</i> , 2013, 14, 65.	1.2	8
45	Hi-Plex for high-throughput mutation screening: application to the breast cancer susceptibility gene PALB2. <i>BMC Medical Genomics</i> , 2013, 6, 48.	0.7	13
46	Cross-platform compatibility of Hi-Plex, a streamlined approach for targeted massively parallel sequencing. <i>Analytical Biochemistry</i> , 2013, 442, 127-129.	1.1	11
47	MYRF Is a Membrane-Associated Transcription Factor That Autoproteolytically Cleaves to Directly Activate Myelin Genes. <i>PLoS Biology</i> , 2013, 11, e1001625.	2.6	198
48	A high-plex PCR approach for massively parallel sequencing. <i>BioTechniques</i> , 2013, 55, 69-74.	0.8	51
49	Bpipe: a tool for running and managing bioinformatics pipelines. <i>Bioinformatics</i> , 2012, 28, 1525-1526.	1.8	145
50	Performance of Hybrid Programming Models for Multiscale Cardiac Simulations: Preparing for Petascale Computation. <i>IEEE Transactions on Biomedical Engineering</i> , 2011, 58, 2965-2969.	2.5	15
51	Petascale computation performance of lightweight multiscale cardiac models using hybrid programming models. , 2011, 2011, 433-6.		1
52	A lightweight interactive debugger for haskell. , 2007, , .		14
53	Practical aspects of declarative debugging in Haskell 98. , 2003, , .		20
54	Specialisation of Higher-Order Functions for Debugging. <i>Electronic Notes in Theoretical Computer Science</i> , 2002, 64, 277-291.	0.9	1