Bernard J Pope

List of Publications by Year
in descending order

54	2,218	471509 17	254184 43
papers	citations	h-index	g-index

62	62	62	4831
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Long-read assembly and comparative evidence-based reanalysis of Cryptosporidium genome sequences reveal expanded transporter repertoire and duplication of entire chromosome ends including subtelomeric regions. Genome Research, 2022, 32, 203-213.	5.5	26
2	Phase 2 Study of Neoadjuvant FGFR Inhibition and Androgen Deprivation Therapy Prior to Prostatectomy. Clinical Genitourinary Cancer, 2022, 20, 452-458.	1.9	5
3	Identifying colorectal cancer caused by biallelic MUTYH pathogenic variants using tumor mutational signatures. Nature Communications, 2022, 13, .	12.8	15
4	Evaluating the utility of tumour mutational signatures for identifying hereditary colorectal cancer and polyposis syndrome carriers. Gut, 2021, 70, 2138-2149.	12.1	27
5	Germline and Tumor Sequencing as a Diagnostic Tool To Resolve Suspected Lynch Syndrome. Journal of Molecular Diagnostics, 2021, 23, 358-371.	2.8	12
6	MSH2-deficient prostate tumours have a distinct immune response and clinical outcome compared to MSH2-deficient colorectal or endometrial cancer. Prostate Cancer and Prostatic Diseases, 2021, 24, 1167-1180.	3.9	4
7	Rare germline variants in the AXIN2 gene in families with colonic polyposis and colorectal cancer. Familial Cancer, 2021, , 1.	1.9	5
8	Population-based estimates of breast cancer risk for carriers of pathogenic variants identified by gene-panel testing. Npj Breast Cancer, 2021, 7, 153.	5.2	10
9	SNPPar: identifying convergent evolution and other homoplasies from microbial whole-genome alignments. Microbial Genomics, 2021, 7, .	2.0	5
10	HiTIME: An efficient model-selection approach for the detection of unknown drug metabolites in LC-MS data. SoftwareX, 2020, 12, 100559.	2.6	3
11	Monoallelic NTHL1 Loss-of-Function Variants and Risk of Polyposis and Colorectal Cancer. Gastroenterology, 2020, 159, 2241-2243.e6.	1.3	20
12	Genetic testing in Poland and Ukraine: should comprehensive germline testing of BRCA1 and BRCA2 be recommended for women with breast and ovarian cancer?. Genetical Research, 2020, 102, e6.	0.9	12
13	Detection of ctDNA in plasma of patients with clinically localised prostate cancer is associated with rapid disease progression. Genome Medicine, 2020, 12, 72.	8.2	35
14	Mismatch repair gene pathogenic germline variants in a population-based cohort of breast cancer. Familial Cancer, 2020, 19, 197-202.	1.9	6
15	Rare germline genetic variants and risk of aggressive prostate cancer. International Journal of Cancer, 2020, 147, 2142-2149.	5.1	12
16	Hi-Plex2: a simple and robust approach to targeted sequencing-based genetic screening. BioTechniques, 2019, 67, 118-122.	1.8	11
17	Bionitio: demonstrating and facilitating best practices for bioinformatics command-line software. GigaScience, 2019, 8, .	6.4	13
18	Tumor mutational signatures in sebaceous skin lesions from individuals with Lynch syndrome. Molecular Genetics & Genomic Medicine, 2019, 7, e00781.	1.2	8

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19	Annotation of theGiardiaproteome through structure-based homology and machine learning. GigaScience, 2019, 8, .	6.4	21
20	Is RNASEL:p.Glu265* a modifier of early-onset breast cancer risk for carriers of high-risk mutations?. BMC Cancer, 2018, 18, 165.	2.6	6
21	Somatic mutations of the coding microsatellites within the beta-2-microglobulin gene in mismatch repair-deficient colorectal cancers and adenomas. Familial Cancer, 2018, 17, 91-100.	1.9	21
22	Targeted massively parallel sequencing characterises the mutation spectrum of PALB2 in breast and ovarian cancer cases from Poland and Ukraine. Familial Cancer, 2018, 17, 345-349.	1.9	7
23	Hi-Plex for Simple, Accurate, and Cost-Effective Amplicon-based Targeted DNA Sequencing. Methods in Molecular Biology, 2018, 1712, 53-70.	0.9	2
24	Risk of colorectal cancer for carriers of a germ-line mutation in POLE or POLD1. Genetics in Medicine, 2018, 20, 890-895.	2.4	49
25	sEst: Accurate Sex-Estimation and Abnormality Detection in Methylation Microarray Data. International Journal of Molecular Sciences, 2018, 19, 3172.	4.1	8
26	FANCM and RECQL genetic variants and breast cancer susceptibility: relevance to South Poland and West Ukraine. BMC Medical Genetics, 2018, 19, 12.	2.1	20
27	Mutation screening of ACKR3 and COPS8 in kidney cancer cases from the CONFIRM study. Familial Cancer, 2017, 16, 411-416.	1.9	6
28	Single nucleotide-level mapping of DNA double-strand breaks in human HEK293T cells. Genomics Data, 2017, 11, 43-45.	1.3	0
29	Variant effect prediction tools assessed using independent, functional assay-based datasets: implications for discovery and diagnostics. Human Genomics, 2017, 11, 10.	2.9	68
30	A novel Drosophila injury model reveals severed axons are cleared through a Draper/MMP-1 signaling cascade. ELife, 2017, 6, .	6.0	47
31	Four simple recommendations to encourage best practices in research software. F1000Research, 2017, 6, 876.	1.6	88
32	Best practice data life cycle approaches for the life sciences. F1000Research, 2017, 6, 1618.	1.6	21
33	Best practice data life cycle approaches for the life sciences. F1000Research, 2017, 6, 1618.	1.6	23
34	Fine resolution mapping of double-strand break sites for human ribosomal DNA units. Genomics Data, 2016, 10, 19-21.	1.3	1
35	UNDR ROVER - a fast and accurate variant caller for targeted DNA sequencing. BMC Bioinformatics, 2016, 17, 165.	2.6	3
36	MethPat: a tool for the analysis and visualisation of complex methylation patterns obtained by massively parallel sequencing. BMC Bioinformatics, 2016, 17, 98.	2.6	22

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37	Exemplary multiplex bisulfite amplicon data used to demonstrate the utility of Methpat. GigaScience, 2015, 4, 55.	6.4	3
38	Abridged adapter primers increase the target scope of Hi-Plex. BioTechniques, 2015, 58, 33-6.	1.8	7
39	Mutation screening of PALB2 in clinically ascertained families from the Breast Cancer Family Registry. Breast Cancer Research and Treatment, 2015, 149, 547-554.	2.5	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. Analytical Chemistry, 2015, 87, 4104-4109.	6.5	23
41	SRST2: Rapid genomic surveillance for public health and hospital microbiology labs. Genome Medicine, 2014, 6, 90.	8.2	953
42	ROVER variant caller: read-pair overlap considerate variant-calling software applied to PCR-based massively parallel sequencing datasets. Source Code for Biology and Medicine, 2014, 9, 3.	1.7	12
43	Rare Mutations in RINT1 Predispose Carriers to Breast and Lynch Syndromeâ€“Spectrum Cancers. Cancer Discovery, 2014, 4, 804-815.	9.4	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. BMC Bioinformatics, 2013, 14, 65.	2.6	8
45	Hi-Plex for high-throughput mutation screening: application to the breast cancer susceptibility gene PALB2. BMC Medical Genomics, 2013, 6, 48.	1.5	13
46	Cross-platform compatibility of Hi-Plex, a streamlined approach for targeted massively parallel sequencing. Analytical Biochemistry, 2013, 442, 127-129.	2.4	11
47	MYRF Is a Membrane-Associated Transcription Factor That Autoproteolytically Cleaves to Directly Activate Myelin Genes. PLoS Biology, 2013, 11, e1001625.	5.6	198
48	A high-plex PCR approach for massively parallel sequencing. BioTechniques, 2013, 55, 69-74.	1.8	51
49	Bpipe: a tool for running and managing bioinformatics pipelines. Bioinformatics, 2012, 28, 1525-1526.	4.1	145
50	Performance of Hybrid Programming Models for Multiscale Cardiac Simulations: Preparing for Petascale Computation. IEEE Transactions on Biomedical Engineering, 2011, 58, 2965-2969.	4.2	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. Electronic Notes in Theoretical Computer Science, 2002, 64, 277-291.	0.9	1