Paul A James

List of Publications by Year
in descending order

95 papers	4,876 citations	117625 34 h-index	102487 66 g-index
97 all docs	97 docs citations	97 times ranked	9308 citing authors

#	Article	IF	Citations
1	Breast-Cancer Risk in Families with Mutations in PALB2. New England Journal of Medicine, 2014, 371, 497-506.	27.0	745
2	A prospective evaluation of whole-exome sequencing as a first-tier molecular test in infants with suspected monogenic disorders. Genetics in Medicine, 2016, 18, 1090-1096.	2.4	332
3	Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. PLoS Genetics, 2013, 9, e1003212.	3.5	244
4	Cancer Risks forMLH1andMSH2Mutation Carriers. Human Mutation, 2013, 34, 490-497.	2.5	201
5	Exome Sequencing Identifies Rare Deleterious Mutations in DNA Repair Genes FANCC and BLM as Potential Breast Cancer Susceptibility Alleles. PLoS Genetics, 2012, 8, e1002894.	3.5	186
6	Monogenic and polygenic determinants of sarcoma risk: an international genetic study. Lancet Oncology, The, 2016, 17, 1261-1271.	10.7	161
7	Panel Testing for Familial Breast Cancer: Calibrating the Tension Between Research and Clinical Care. Journal of Clinical Oncology, 2016, 34, 1455-1459.	1.6	154
8	A mutation in the small heat-shock protein HSPB1 leading to distal hereditary motor neuronopathy disrupts neurofilament assembly and the axonal transport of specific cellular cargoes. Human Molecular Genetics, 2006, 15, 347-354.	2.9	138
9	Mutational Signature Analysis Reveals NTHL1 Deficiency to Cause a Multi-tumor Phenotype. Cancer Cell, 2019, 35, 256-266.e5.	16.8	123
10	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	21.4	120
11	Optimal Selection of Individuals for BRCA Mutation Testing: A Comparison of Available Methods. Journal of Clinical Oncology, 2006, 24, 707-715.	1.6	112
12	Large scale multifactorial likelihood quantitative analysis of BRCA1 and BRCA2 variants: An ENIGMA resource to support clinical variant classification. Human Mutation, 2019, 40, 1557-1578.	2.5	102
13	Clinical applications of polygenic breast cancer risk: a critical review and perspectives of an emerging field. Breast Cancer Research, 2020, 22, 21.	5.0	98
14	No evidence that protein truncating variants inBRIP1are associated with breast cancer risk: implications for gene panel testing. Journal of Medical Genetics, 2016, 53, 298-309.	3.2	94
15	Real-world utility of whole exome sequencing with targeted gene analysis for focal epilepsy. Epilepsy Research, 2017, 131, 1-8.	1.6	93
16	FANCMc.5791C>T nonsense mutation (rs144567652) induces exon skipping, affects DNA repair activity and is a familial breast cancer risk factor. Human Molecular Genetics, 2015, 24, 5345-5355.	2.9	91
17	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. Genetics in Medicine, 2020, 22, 1653-1666.	2.4	82
18	Specifications of the ACMG/AMP variant interpretation guidelines for germline TP53 variants. Human Mutation, 2021, 42, 223-236.	2.5	81

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19	Cpipe: a shared variant detection pipeline designed for diagnostic settings. Genome Medicine, 2015, 7, 68.	8.2	78
20	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. Journal of the National Cancer Institute, 2016, 108, djv315.	6.3	77
21	A Role for Common Genomic Variants in the Assessment of Familial Breast Cancer. Journal of Clinical Oncology, 2012, 30, 4330-4336.	1.6	74
22	Analysis of RAD51C germline mutations in high-risk breast and ovarian cancer families and ovarian cancer patients. Human Mutation, 2012, 33, 95-99.	2.5	64
23	Population genomic screening of all young adults in a health-care system: a cost-effectiveness analysis. Genetics in Medicine, 2019, 21, 1958-1968.	2.4	63
24	Variation in the risk of colorectal cancer in families with Lynch syndrome: a retrospective cohort study. Lancet Oncology, The, 2021, 22, 1014-1022.	10.7	58
25	Breast cancer risk prediction using a polygenic risk score in the familial setting: a prospective study from the Breast Cancer Family Registry and kConFab. Genetics in Medicine, 2017, 19, 30-35.	2.4	53
26	Rare variants in XRCC2 as breast cancer susceptibility alleles: TableÂ1. Journal of Medical Genetics, 2012, 49, 618-620.	3.2	49
27	ALPK3-deficient cardiomyocytes generated from patient-derived induced pluripotent stem cells and mutant human embryonic stem cells display abnormal calcium handling and establish that ALPK3 deficiency underlies familial cardiomyopathy. European Heart Journal, 2016, 37, 2586-2590.	2.2	49
28	Characterization of the Cancer Spectrum in Men With GermlineBRCA1andBRCA2Pathogenic Variants. JAMA Oncology, 2020, 6, 1218.	7.1	48
29	Molecular analysis of PALB2â€associated breast cancers. Journal of Pathology, 2018, 245, 53-60.	4.5	46
30	Population-based genetic testing of asymptomatic women for breast and ovarian cancer susceptibility. Genetics in Medicine, 2019, 21, 913-922.	2.4	45
31	The molecular genetics of non-ALS motor neuron diseases. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2006, 1762, 986-1000.	3.8	41
32	Extending the scope of diagnostic chromosome analysis: Detection of single gene defects using high-resolution SNP microarrays. Human Mutation, 2011, 32, 1500-1506.	2.5	41
33	Analysis of RAD51D in Ovarian Cancer Patients and Families with a History of Ovarian or Breast Cancer. PLoS ONE, 2013, 8, e54772.	2.5	36
34	Prevalence of PALB2 mutations in Australian familial breast cancer cases and controls. Breast Cancer Research, 2015, 17, 111.	5.0	36
35	Diagnostic and cost utility of whole exome sequencing in peripheral neuropathy. Annals of Clinical and Translational Neurology, 2017, 4, 318-325.	3.7	36
36	Current review ofTP53pathogenic germline variants in breast cancer patients outside Li-Fraumeni syndrome. Human Mutation, 2018, 39, 1764-1773.	2.5	35

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37	Improved, ACMG-compliant, in silico prediction of pathogenicity for missense substitutions encoded by TP53 variants. Human Mutation, 2018, 39, 1061-1069.	2.5	29
38	Cost-effectiveness and comparative effectiveness of cancer risk management strategies in BRCA1/2 mutation carriers: a systematic review. Genetics in Medicine, 2018, 20, 1145-1156.	2.4	28
39	Mainstream genetic testing for breast cancer patients: early experiences from the Parkville Familial Cancer Centre. European Journal of Human Genetics, 2021, 29, 872-880.	2.8	27
40	The Angelina Jolie effect. Medical Journal of Australia, 2013, 199, 646-646.	1.7	26
41	Reevaluation of the BRCA2 truncating allele c.9976Aâ€‰>â€‰T (p.Lys3326Ter) in a familial breast cancer context. Scientific Reports, 2015, 5, 14800.	3.3	26
42	Combined Tumor Sequencing and Case-Control Analyses of RAD51C in Breast Cancer. Journal of the National Cancer Institute, 2019, 111, 1332-1338.	6.3	26
43	Expanding the clinical and genetic spectrum of ALPK3 variants: Phenotypes identified in pediatric cardiomyopathy patients and adults with heterozygous variants. American Heart Journal, 2020, 225, 108-119.	2.7	25
44	A clinically driven variant prioritization framework outperforms purely computational approaches for the diagnostic analysis of singleton WES data. European Journal of Human Genetics, 2017, 25, 1268-1272.	2.8	24
45	Exome sequencing of familial high-grade serous ovarian carcinoma reveals heterogeneity for rare candidate susceptibility genes. Nature Communications, 2020, 11, 1640.	12.8	24
46	Polygenic risk modeling for prediction of epithelial ovarian cancer risk. European Journal of Human Genetics, 2022, 30, 349-362.	2.8	23
47	Large genomic rearrangements in the familial breast and ovarian cancer gene BRCA1 are associated with an increased frequency of high risk features. Familial Cancer, 2015, 14, 287-295.	1.9	22
48	A quantitative model to predict pathogenicity of missense variants in the TP53 gene. Human Mutation, 2019, 40, 788-800.	2.5	21
49	Evaluation of telephone genetic counselling to facilitate germline BRCA1/2 testing in women with high-grade serous ovarian cancer. European Journal of Human Genetics, 2019, 27, 1186-1196.	2.8	21
50	Medically actionable pathogenic variants in a population of 13,131 healthy elderly individuals. Genetics in Medicine, 2020, 22, 1883-1886.	2.4	20
51	Evaluating the breast cancer predisposition role of rare variants in genes associated with low-penetrance breast cancer risk SNPs. Breast Cancer Research, 2018, 20, 3.	5.0	19
52	Mutations in RECQL are not associated with breast cancer risk in an Australian population. Nature Genetics, 2018, 50, 1346-1348.	21.4	19
53	Uptake of polygenic risk information among women at increased risk of breast cancer. Clinical Genetics, 2020, 97, 492-501.	2.0	19
54	Cost-effectiveness of long-term clinical management of BRCA pathogenic variant carriers. Genetics in Medicine, 2020, 22, 831-839.	2.4	19

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55	A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. Nature Communications, 2021, 12, 1078.	12.8	19
56	Breast and Prostate Cancer Risks for MaleBRCA1andBRCA2Pathogenic Variant Carriers Using Polygenic Risk Scores. Journal of the National Cancer Institute, 2022, 114, 109-122.	6.3	19
57	Clinical implications of genomics for cancer risk genetics. Lancet Oncology, The, 2015, 16, e303-e308.	10.7	17
58	The Cardiac Genetics Clinic: a model for multidisciplinary genomic medicine. Medical Journal of Australia, 2015, 203, 261-261.	1.7	16
59	Reevaluation of RINT1 as a breast cancer predisposition gene. Breast Cancer Research and Treatment, 2016, 159, 385-392.	2.5	16
60	Suggested application of HER2+ breast tumor phenotype for germline TP53 variant classification within ACMG/AMP guidelines. Human Mutation, 2020, 41, 1555-1562.	2.5	16
61	The clinical utility of exome sequencing and extended bioinformatic analyses in adolescents and adults with a broad range of neurological phenotypes: an Australian perspective. Journal of the Neurological Sciences, 2021, 420, 117260.	0.6	16
62	The predictive ability of the 313 variantâ€“based polygenic risk score for contralateral breast cancer risk prediction in women of European ancestry with a heterozygous BRCA1 or BRCA2 pathogenic variant. Genetics in Medicine, 2021, 23, 1726-1737.	2.4	16
63	Molecular comparison of interval and screenâ€detected breast cancers. Journal of Pathology, 2019, 248, 243-252.	4.5	15
64	A novel approach to offering additional genomic findingsâ€”A protocol to test a twoâ€step approach in the healthcare system. Journal of Genetic Counseling, 2019, 28, 388-397.	1.6	14
65	Breast cancer polygenic risk scores: a 12-month prospective study of patient reported outcomes and risk management behavior. Genetics in Medicine, 2021, 23, 2316-2323.	2.4	14
66	Mapping the EORTC-QLQ-C30 to the EQ-5D-3L: An Assessment of Existing and Newly Developed Algorithms. Medical Decision Making, 2018, 38, 954-967.	2.4	13
67	Influence of lived experience on risk perception among women who received a breast cancer polygenic risk score: â€˜Another piece of the pieâ€™. Journal of Genetic Counseling, 2021, 30, 849-860.	1.6	13
68	Communicating polygenic risk scores in the familial breast cancer clinic. Patient Education and Counseling, 2021, 104, 2512-2521.	2.2	12
69	Investigation of monogenic causes of familial breast cancer: data from the BEACCON case-control study. Npj Breast Cancer, 2021, 7, 76.	5.2	12
70	A functionally impaired missense variant identified in French Canadian families implicates FANCI as a candidate ovarian cancer-predisposing gene. Genome Medicine, 2021, 13, 186.	8.2	12
71	The incidence of PALB2 c.3113G>A in women with a strong family history of breast and ovarian cancer attending familial cancer centres in Australia. Familial Cancer, 2013, 12, 587-595.	1.9	11
72	p53 major hotspot variants are associated with poorer prognostic features in hereditary cancer patients. Cancer Genetics, 2019, 235-236, 21-27.	0.4	11

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73	Genetic variants associated with inherited cardiovascular disorders among 13,131 asymptomatic older adults of European descent. Npj Genomic Medicine, 2021, 6, 51.	3.8	11
74	Risks of breast and ovarian cancer for women harboring pathogenic missense variants in BRCA1 and BRCA2 compared with those harboring protein truncating variants. Genetics in Medicine, 2022, 24, 119-129.	2.4	10
75	â€œCancer 2015â€• A Prospective, Population-Based Cancer Cohortâ€”Phase 1: Feasibility of Genomics-Guided Precision Medicine in the Clinic. Journal of Personalized Medicine, 2015, 5, 354-369.	2.5	8
76	A Microsimulation Model for Evaluating the Effectiveness of Cancer Risk Management for BRCA Pathogenic Variant Carriers: miBRovaCAre. Value in Health, 2019, 22, 854-862.	0.3	8
77	Development and pilot testing of a leaflet informing women with breast cancer about genomic testing for polygenic risk. Familial Cancer, 2019, 18, 147-152.	1.9	8
78	Polygenic score modifies risk for Alzheimer's disease in APOE Îµ4 homozygotes at phenotypic extremes. Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring, 2021, 13, e12226.	2.4	7
79	Evaluation of the association of heterozygous germline variants in NTHL1 with breast cancer predisposition: an international multi-center study of 47,180 subjects. Npj Breast Cancer, 2021, 7, 52.	5.2	7
80	An updated quantitative model to classify missense variants in the TP53 gene: A novel multifactorial strategy. Human Mutation, 2021, 42, 1351-1361.	2.5	7
81	A rapid scoring tool to assess mutation probability in patients with inherited cardiac disorders. European Journal of Medical Genetics, 2018, 61, 61-67.	1.3	6
82	Changing landscape of hereditary breast and ovarian cancer germline genetic testing in Australia. Internal Medicine Journal, 2018, 48, 1269-1272.	0.8	6
83	Cancer risk management in Tasmanian women with BRCA1 and BRCA2 mutations. Familial Cancer, 2018, 17, 333-344.	1.9	5
84	Differences in patient ascertainment affect the use of geneâ€specified ACMG/AMP phenotypeâ€related variant classification criteria: Evidence for TP53. Human Mutation, 2020, 41, 537-542.	2.5	5
85	Polygenic risk in familial breast cancer: Changing the dynamics of communicating genetic risk. Journal of Genetic Counseling, 2022, 31, 120-129.	1.6	4
86	Suspected clonal hematopoiesis as a natural functional assay of TP53 germline variant pathogenicity. Genetics in Medicine, 2022, 24, 673-680.	2.4	4
87	Real world outcomes and implementation pathways of exome sequencing in an adult genetic department. Genetics in Medicine, 2022, , .	2.4	4
88	Caseâ€“case analysis addressing ascertainment bias for multigene panel testing implicates BRCA1 and PALB2 in endometrial cancer. Human Mutation, 2021, 42, 1265-1278.	2.5	3
89	The Clinical and Psychosocial Outcomes for Women Who Received Unexpected Clinically Actionable Germline Information Identified through Research: An Exploratory Sequential Mixed-Methods Comparative Study. Journal of Personalized Medicine, 2022, 12, 1112.	2.5	2
90	Mutational Signature Analysis Reveals NTHL1 Deficiency to Cause a Multi-Tumor Phenotype Including a Predisposition to Colon and Breast Cancer. SSRN Electronic Journal, 0, , .	0.4	1

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91	Universal genetic testing of patients with newly diagnosed breast cancer â€" ready for prime time?. Medical Journal of Australia, 2021, 215, 449-453.	1.7	1
92	Contribution of large genomic rearrangements inPALB2to familial breast cancer: implications for genetic testing. Journal of Medical Genetics, 2023, 60, 112-118.	3.2	1
93	Unselected Women's Experiences of Receiving Genetic Research Results for Hereditary Breast and Ovarian Cancer: A Qualitative Study. Genetic Testing and Molecular Biomarkers, 2021, 25, 741-748.	0.7	1
94	Oncologistsâ€™ perspectives of telephone genetic counseling to facilitate germline BRCA1/2 testing for their patients with high-grade serous ovarian cancer. Journal of Community Genetics, 2021, 12, 449-457.	1.2	0
95	Integration of tumour sequencing and caseâ€“control data to assess pathogenicity of RAD51C missense variants in familial breast cancer. Npj Breast Cancer, 2022, 8, 10.	5.2	0