Paul James

List of Publications by Year
in descending order

212 papers	10,055 citations	43973 48 h-index	46693 89 g-index
229 all docs	229 docs citations	229 times ranked	15923 citing authors

#	Article	IF	Citations
1	Contribution of large genomic rearrangements inPALB2to familial breast cancer: implications for genetic testing. Journal of Medical Genetics, 2023, 60, 112-118.	1.5	1
2	Population-basedBRCA1/2testing programmes are highly acceptable in the Jewish community: results of the JeneScreen Study. Journal of Medical Genetics, 2023, 60, 265-273.	1.5	4
3	Metaphors and why these are important in all aspects of genetic counseling. Journal of Genetic Counseling, 2022, 31, 34-40.	0.9	4
4	Polygenic risk in familial breast cancer: Changing the dynamics of communicating genetic risk. Journal of Genetic Counseling, 2022, 31, 120-129.	0.9	4
5	Breast and Prostate Cancer Risks for MaleBRCA1andBRCA2Pathogenic Variant Carriers Using Polygenic Risk Scores. Journal of the National Cancer Institute, 2022, 114, 109-122.	3.0	19
6	Stakeholder attitudes towards establishing a national genomics registry of inherited cancer predisposition: a qualitative study. Journal of Community Genetics, 2022, 13, 59-73.	0.5	2
7	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. Genetics in Medicine, 2022, 24, 130-145.	1.1	45
8	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.	1.1	10
9	Suspected clonal hematopoiesis as a natural functional assay of TP53 germline variant pathogenicity. Genetics in Medicine, 2022, 24, 673-680.	1.1	4
10	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.	2.3	0
11	Polygenic risk modeling for prediction of epithelial ovarian cancer risk. European Journal of Human Genetics, 2022, 30, 349-362.	1.4	23
12	Predictors and outcomes of in-hospital referrals for forensic investigation after young sudden cardiac death. Heart Rhythm, 2022, 19, 937-944.	0.3	8
13	Pregnancy Outcomes in Females With Dilated Cardiomyopathyâ€“Associated Rare Genetic Variants. Circulation Genomic and Precision Medicine, 2022, , CIRCGEN121003540.	1.6	0
14	Arrhythmic Phenotypes Are a Defining Feature of Dilated Cardiomyopathy-Associated SCN5A Variants: A Systematic Review. Circulation Genomic and Precision Medicine, 2022, 15, CIRCGEN121003432.	1.6	13
15	Estimating the proportion of pathogenic variants from breast cancer caseâ€“control data: Application to calibration of ACMG/AMP variant classification criteria. Human Mutation, 2022, 43, 882-888.	1.1	6
16	TRACEBACK: Testing of Historical Tubo-Ovarian Cancer Patients for Hereditary Risk Genes as a Cancer Prevention Strategy in Family Members. Journal of Clinical Oncology, 2022, , JCO2102108.	0.8	3
17	Population DNA screening for medically actionable disease risk in adults. Medical Journal of Australia, 2022, 216, 278-280.	0.8	10
18	Real world outcomes and implementation pathways of exome sequencing in an adult genetic department. Genetics in Medicine, 2022, , .	1.1	4

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19	Prevalence of Coronary Artery Anomalies in Young and Middle-Aged Sudden Cardiac Death Victims (from a Prospective State-Wide Registry). American Journal of Cardiology, 2022, , .	0.7	1
20	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.	1.1	2
21	Implementing gene curation for hereditary cancer susceptibility in Australia: achieving consensus on genes with clinical utility. Journal of Medical Genetics, 2021, 58, 853-858.	1.5	3
22	Finding the fiveâ€year window: A qualitative study examining young women's decisionâ€making and experience of using tamoxifen to reduce BRCA1/2 breast cancer risk. Psycho-Oncology, 2021, 30, 159-166.	1.0	3
23	Common Susceptibility Loci for Male Breast Cancer. Journal of the National Cancer Institute, 2021, 113, 453-461.	3.0	12
24	The End Unexplained Cardiac Death (EndUCD) Registry for Young Australian Sudden Cardiac Arrest. Heart Lung and Circulation, 2021, 30, 714-720.	0.2	18
25	Specifications of the ACMG/AMP variant interpretation guidelines for germline TP53 variants. Human Mutation, 2021, 42, 223-236.	1.1	81
26	Can post-mortem coronary artery calcium scores aid diagnosis in young sudden death?. Forensic Science, Medicine, and Pathology, 2021, 17, 27-35.	0.6	2
27	Globalization in question: why does engaged theory matter?. Globalizations, 2021, 18, 794-809.	1.9	3
28	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.	1.2	7
29	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.	0.3	16
30	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.	0.9	13
31	Clinical and laboratory reporting impact of ACMG-AMP and modified ClinGen variant classification frameworks in MYH7-related cardiomyopathy. Genetics in Medicine, 2021, 23, 1108-1115.	1.1	14
32	A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. Nature Communications, 2021, 12, 1078.	5.8	19
33	Genetic testing in dementiaâ€A medical genetics perspective. International Journal of Geriatric Psychiatry, 2021, 36, 1158-1170.	1.3	9
34	Communicating polygenic risk scores in the familial breast cancer clinic. Patient Education and Counseling, 2021, 104, 2512-2521.	1.0	12
35	Mainstream genetic testing for breast cancer patients: early experiences from the Parkville Familial Cancer Centre. European Journal of Human Genetics, 2021, 29, 872-880.	1.4	27
36	Management of individuals with germline variants in PALB2: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 1416-1423.	1.1	34

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37	Cancer Diagnoses Following Abnormal Noninvasive Prenatal Testing: A Case Series, Literature Review, and Proposed Management Model. JCO Precision Oncology, 2021, 5, 1001-1012.	1.5	19
38	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.	2.3	7
39	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.	0.5	0
40	Genetic Dominant Variants in STUB1, Segregating in Families with SCA48, Display In Vitro Functional Impairments Indistinctive from Recessive Variants Associated with SCAR16. International Journal of Molecular Sciences, 2021, 22, 5870.	1.8	10
41	The economic impact of sudden cardiac arrest. Resuscitation, 2021, 163, 49-56.	1.3	13
42	Evaluation of two population screening programmes for BRCA1/2 founder mutations in the Australian Jewish community: a protocol paper. BMJ Open, 2021, 11, e041186.	0.8	2
43	Investigation of monogenic causes of familial breast cancer: data from the BEACCON case-control study. Npj Breast Cancer, 2021, 7, 76.	2.3	12
44	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.	1.1	16
45	Genetic variants associated with inherited cardiovascular disorders among 13,131 asymptomatic older adults of European descent. Npj Genomic Medicine, 2021, 6, 51.	1.7	11
46	Genomic Risk Prediction for Breast Cancer in Older Women. Cancers, 2021, 13, 3533.	1.7	6
47	Caseâ€“case analysis addressing ascertainment bias for multigene panel testing implicates BRCA1 and PALB2 in endometrial cancer. Human Mutation, 2021, 42, 1265-1278.	1.1	3
48	Variation in the risk of colorectal cancer in families with Lynch syndrome: a retrospective cohort study. Lancet Oncology, The, 2021, 22, 1014-1022.	5.1	58
49	An updated quantitative model to classify missense variants in the TP53 gene: A novel multifactorial strategy. Human Mutation, 2021, 42, 1351-1361.	1.1	7
50	Mendelian randomisation study of smoking exposure in relation to breast cancer risk. British Journal of Cancer, 2021, 125, 1135-1145.	2.9	9
51	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.	1.1	14
52	Long-Term Efficacy and Safety of Sodium Channel Antagonists in Patients With p.R222Q SCN5A-Related Arrhythmic Dilated Cardiomyopathy. JACC: Clinical Electrophysiology, 2021, 7, 126-128.	1.3	2
53	A prospective prostate cancer screening programme for men with pathogenic variants in mismatch repair genes (IMPACT): initial results from an international prospective study. Lancet Oncology, The, 2021, 22, 1618-1631.	5.1	48
54	Universal genetic testing of patients with newly diagnosed breast cancer â€” ready for prime time?. Medical Journal of Australia, 2021, 215, 449-453.	0.8	1

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55	Population-based estimates of breast cancer risk for carriers of pathogenic variants identified by gene-panel testing. Npj Breast Cancer, 2021, 7, 153.	2.3	10
56	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.	0.3	1
57	A functionally impaired missense variant identified in French Canadian families implicates FANCI as a candidate ovarian cancer-predisposing gene. Genome Medicine, 2021, 13, 186.	3.6	12
58	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.4	39
59	Management of People With a Fontan Circulation: a Cardiac Society of Australia and New Zealand Position statement. Heart Lung and Circulation, 2020, 29, 5-39.	0.2	42
60	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	9.4	120
61	Cancer Risks Associated With Germline PALB2 Pathogenic Variants: An International Study of 524 Families. Journal of Clinical Oncology, 2020, 38, 674-685.	0.8	270
62	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.	1.1	5
63	Uptake of polygenic risk information among women at increased risk of breast cancer. Clinical Genetics, 2020, 97, 492-501.	1.0	19
64	Genotype-phenotype correlations among TP53 carriers: Literature review and analysis of probands undergoing multi-gene panel testing and single-gene testing. Cancer Genetics, 2020, 248-249, 11-17.	0.2	8
65	Disjunctive Globalization in the Era of the Great Unsettling. Theory, Culture and Society, 2020, 37, 187-203.	1.3	15
66	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.	1.1	82
67	Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. American Journal of Human Genetics, 2020, 107, 837-848.	2.6	39
68	Association of germline variation with the survival of women with BRCA1/2 pathogenic variants and breast cancer. Npj Breast Cancer, 2020, 6, 44.	2.3	5
69	Suicide in frontotemporal dementia and Huntington disease: analysis of family-reported pedigree data and implications for genetic healthcare for asymptomatic relatives. Psychology and Health, 2020, 36, 1-7.	1.2	3
70	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581.	9.4	265
71	The development and evaluation of a nationwide training program for oncology health professionals in the provision of genetic testing for ovarian cancer patients. Gynecologic Oncology, 2020, 158, 431-439.	0.6	15
72	Suggested application of HER2+ breast tumor phenotype for germline TP53 variant classification within ACMG/AMP guidelines. Human Mutation, 2020, 41, 1555-1562.	1.1	16

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73	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.	1.2	25
74	Womenâ€™s responses and understanding of polygenic breast cancer risk information. Familial Cancer, 2020, 19, 297-306.	0.9	15
75	Population Genomic Screening of All Young Adults in a Health-Care System: A Cost-Effectiveness Analysis. Obstetrical and Gynecological Survey, 2020, 75, 91-93.	0.2	2
76	A synonymous GATA2 variant underlying familial myeloid malignancy with striking intrafamilial phenotypic variability. British Journal of Haematology, 2020, 190, e297-e301.	1.2	14
77	Medically actionable pathogenic variants in a population of 13,131 healthy elderly individuals. Genetics in Medicine, 2020, 22, 1883-1886.	1.1	20
78	Characterization of the Cancer Spectrum in Men With GermlineBRCA1andBRCA2Pathogenic Variants. JAMA Oncology, 2020, 6, 1218.	3.4	48
79	Emerging role of genetic analysis for stratification of sudden cardiac death risk in dilated cardiomyopathy: An illustrative case. HeartRhythm Case Reports, 2020, 6, 499-502.	0.2	1
80	Familial Aortopathies â€“ State of the Art Review. Heart Lung and Circulation, 2020, 29, 607-618.	0.2	8
81	Clinical applications of polygenic breast cancer risk: a critical review and perspectives of an emerging field. Breast Cancer Research, 2020, 22, 21.	2.2	98
82	Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€receptor status. Genetic Epidemiology, 2020, 44, 442-468.	0.6	32
83	Prospective Evaluation of the Utility of Whole Exome Sequencing in Dilated Cardiomyopathy. Journal of the American Heart Association, 2020, 9, e013346.	1.6	28
84	The Medical Genome Reference Bank contains whole genome and phenotype data of 2570 healthy elderly. Nature Communications, 2020, 11, 435.	5.8	47
85	Cost-effectiveness of long-term clinical management of BRCA pathogenic variant carriers. Genetics in Medicine, 2020, 22, 831-839.	1.1	19
86	Exome sequencing of familial high-grade serous ovarian carcinoma reveals heterogeneity for rare candidate susceptibility genes. Nature Communications, 2020, 11, 1640.	5.8	24
87	Cardiac arrest and sudden cardiac death registries: a systematic review of global coverage. Open Heart, 2020, 7, e001195.	0.9	52
88	Arrhythmogenic Right Ventricular Cardiomyopathy: A Review of Living and Deceased Probands. Heart Lung and Circulation, 2019, 28, 1034-1041.	0.2	5
89	High-risk womenâ€™s risk perception after receiving personalized polygenic breast cancer risk information. Journal of Community Genetics, 2019, 10, 197-206.	0.5	16
90	p53 major hotspot variants are associated with poorer prognostic features in hereditary cancer patients. Cancer Genetics, 2019, 235-236, 21-27.	0.2	11

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91	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38.	2.3	28
92	A Microsimulation Model for Evaluating the Effectiveness of Cancer Risk Management for BRCA Pathogenic Variant Carriers: miBRovaCAre. Value in Health, 2019, 22, 854-862.	0.1	8
93	Genetic resilience to Alzheimer's disease in APOE Îµ4 homozygotes: A systematic review. Alzheimer's and Dementia, 2019, 15, 1612-1623.	0.4	21
94	Interim Results from the IMPACT Study: Evidence for Prostate-specific Antigen Screening in BRCA2 Mutation Carriers. European Urology, 2019, 76, 831-842.	0.9	148
95	Shared heritability and functional enrichment across six solid cancers. Nature Communications, 2019, 10, 431.	5.8	88
96	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.	1.1	102
97	The spectrum of BRCA1 and BRCA2 pathogenic sequence variants in Middle Eastern, North African, and South European countries. Human Mutation, 2019, 40, e1-e23.	1.1	34
98	A cost-effectiveness model of genetic testing and periodical clinical screening for the evaluation of families with dilated cardiomyopathy. Genetics in Medicine, 2019, 21, 2815-2822.	1.1	35
99	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	5.8	90
100	A quantitative model to predict pathogenicity of missense variants in the TP53 gene. Human Mutation, 2019, 40, 788-800.	1.1	21
101	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.	1.4	21
102	Higher risk of phaeochromocytoma/paraganglioma (Phaeoâ€Pgl) in SDHD than SDHB carriers: an Australian cohort study. Internal Medicine Journal, 2019, 49, 529-532.	0.5	1
103	Men's experiences of recontact about a potential increased risk of prostate cancer due to Lynch Syndrome: â€œJust another straw on the stackâ€. Journal of Genetic Counseling, 2019, 28, 750-759.	0.9	3
104	Combined Tumor Sequencing and Case-Control Analyses of RAD51C in Breast Cancer. Journal of the National Cancer Institute, 2019, 111, 1332-1338.	3.0	26
105	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.	0.9	14
106	Molecular comparison of interval and screenâ€detected breast cancers. Journal of Pathology, 2019, 248, 243-252.	2.1	15
107	Mutational Signature Analysis Reveals NTHL1 Deficiency to Cause a Multi-tumor Phenotype. Cancer Cell, 2019, 35, 256-266.e5.	7.7	123
108	Population genomic screening of all young adults in a health-care system: a cost-effectiveness analysis. Genetics in Medicine, 2019, 21, 1958-1968.	1.1	63

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109	Insights into sudden cardiac death: exploring the potential relevance of non-diagnostic autopsy findings. European Heart Journal, 2019, 40, 831-838.	1.0	33
110	Development and pilot testing of a leaflet informing women with breast cancer about genomic testing for polygenic risk. Familial Cancer, 2019, 18, 147-152.	0.9	8
111	Population-based genetic testing of asymptomatic women for breast and ovarian cancer susceptibility. Genetics in Medicine, 2019, 21, 913-922.	1.1	45
112	A comparison of Australian and French families affected by sarcoma: perceptions of genetics and incidental findings. Personalized Medicine, 2018, 15, 13-24.	0.8	0
113	The Landscape of Somatic Genetic Alterations in Breast Cancers From ATM Germline Mutation Carriers. Journal of the National Cancer Institute, 2018, 110, 1030-1034.	3.0	90
114	Exploring the feasibility and utility of exomeâ€scale tumour sequencing in a clinical setting. Internal Medicine Journal, 2018, 48, 786-794.	0.5	6
115	Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620.	1.1	224
116	Molecular analysis of PALB2â€associated breast cancers. Journal of Pathology, 2018, 245, 53-60.	2.1	46
117	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.	1.1	28
118	Prostate-specific antigen velocity in a prospective prostate cancer screening study of men with genetic predisposition. British Journal of Cancer, 2018, 118, 266-276.	2.9	12
119	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.	2.2	19
120	Cancer risk management in Tasmanian women with BRCA1 and BRCA2 mutations. Familial Cancer, 2018, 17, 333-344.	0.9	5
121	Making Sense of SNPs: Women's Understanding and Experiences of Receiving a Personalized Profile of Their Breast Cancer Risks. Journal of Genetic Counseling, 2018, 27, 702-708.	0.9	30
122	A rapid scoring tool to assess mutation probability in patients with inherited cardiac disorders. European Journal of Medical Genetics, 2018, 61, 61-67.	0.7	6
123	Prospective validation of the NCI Breast Cancer Risk Assessment Tool (Gail Model) on 40,000 Australian women. Breast Cancer Research, 2018, 20, 155.	2.2	22
124	Changing landscape of hereditary breast and ovarian cancer germline genetic testing in Australia. Internal Medicine Journal, 2018, 48, 1269-1272.	0.5	6
125	Current review ofTP53pathogenic germline variants in breast cancer patients outside Li-Fraumeni syndrome. Human Mutation, 2018, 39, 1764-1773.	1.1	35
126	Mutations in RECQL are not associated with breast cancer risk in an Australian population. Nature Genetics, 2018, 50, 1346-1348.	9.4	19

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127	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.	1.2	13
128	Improved, ACMG-compliant, in silico prediction of pathogenicity for missense substitutions encoded by TP53 variants. Human Mutation, 2018, 39, 1061-1069.	1.1	29
129	Heterogeneity and Uncertainties Specific to Genome-Based Health Technological Assessments. Value in Health, 2018, 21, 891-893.	0.1	1
130	A Transcriptome-Wide Association Study Among 97,898 Women to Identify Candidate Susceptibility Genes for Epithelial Ovarian Cancer Risk. Cancer Research, 2018, 78, 5419-5430.	0.4	54
131	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.	1.1	53
132	Real-world utility of whole exome sequencing with targeted gene analysis for focal epilepsy. Epilepsy Research, 2017, 131, 1-8.	0.8	93
133	Diagnostic and cost utility of whole exome sequencing in peripheral neuropathy. Annals of Clinical and Translational Neurology, 2017, 4, 318-325.	1.7	36
134	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	9.4	356
135	Cousins not twins: intratumoural and intertumoural heterogeneity in syndromic neuroendocrine tumours. Journal of Pathology, 2017, 242, 273-283.	2.1	9
136	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	9.4	289
137	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.	1.4	24
138	Surveillance in Germline TP53 Mutation Carriers Utilizing Whole-Body Magnetic Resonance Imaging. JAMA Oncology, 2017, 3, 1735.	3.4	14
139	Relevance of DNA damage repair in the management of prostate cancer. Current Problems in Cancer, 2017, 41, 287-301.	1.0	16
140	Association of breast cancer risk in BRCA1 and BRCA2 mutation carriers with genetic variants showing differential allelic expression: identification of a modifier of breast cancer risk at locus 11q22.3. Breast Cancer Research and Treatment, 2017, 161, 117-134.	1.1	18
141	Psychosocial and behavioral impact of breast cancer risk assessed by testing for common risk variants: protocol of a prospective study. BMC Cancer, 2017, 17, 491.	1.1	10
142	Prediction of Breast and Prostate Cancer Risks in Male BRCA1 and BRCA2 Mutation Carriers Using Polygenic Risk Scores. Journal of Clinical Oncology, 2017, 35, 2240-2250.	0.8	152
143	1p13.2 deletion displays clinical features overlapping Noonan syndrome, likely related to NRAS gene haploinsufficiency. Genetics and Molecular Biology, 2016, 39, 349-357.	0.6	5
144	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.	1.0	49

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145	Male breast cancer in BRCA1 and BRCA2 mutation carriers: pathology data from the Consortium of Investigators of Modifiers of BRCA1/2. Breast Cancer Research, 2016, 18, 15.	2.2	88
146	Monogenic and polygenic determinants of sarcoma risk: an international genetic study. Lancet Oncology, The, 2016, 17, 1261-1271.	5.1	161
147	Reevaluation of RINT1 as a breast cancer predisposition gene. Breast Cancer Research and Treatment, 2016, 159, 385-392.	1.1	16
148	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	5.8	93
149	A Prospective Study of Sudden Cardiac Death among Children and Young Adults. New England Journal of Medicine, 2016, 374, 2441-2452.	13.9	619
150	Panel Testing for Familial Breast Cancer: Calibrating the Tension Between Research and Clinical Care. Journal of Clinical Oncology, 2016, 34, 1455-1459.	0.8	154
151	Pheo-Type: A Diagnostic Gene-expression Assay for the Classification of Pheochromocytoma and Paraganglioma. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 1034-1043.	1.8	29
152	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.	1.5	94
153	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.	1.1	332
154	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. Journal of the National Cancer Institute, 2016, 108, djv315.	3.0	77
155	Reevaluation of the BRCA2 truncating allele c.9976Aâ€‰>â€‰T (p.Lys3326Ter) in a familial breast cancer context. Scientific Reports, 2015, 5, 14800.	1.6	26
156	Cpipe: a shared variant detection pipeline designed for diagnostic settings. Genome Medicine, 2015, 7, 68.	3.6	78
157	The Cardiac Genetics Clinic: a model for multidisciplinary genomic medicine. Medical Journal of Australia, 2015, 203, 261-261.	0.8	16
158	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.	1.1	8
159	Clinical implications of genomics for cancer risk genetics. Lancet Oncology, The, 2015, 16, e303-e308.	5.1	17
160	Cell-type-specific enrichment of risk-associated regulatory elements at ovarian cancer susceptibility loci. Human Molecular Genetics, 2015, 24, 3595-3607.	1.4	40
161	Copy-number variation associated with congenital anomalies of the kidney and urinary tract. Pediatric Nephrology, 2015, 30, 487-495.	0.9	61
162	The genomic landscape of phaeochromocytoma. Journal of Pathology, 2015, 236, 78-89.	2.1	61

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163	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.	1.4	91
164	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.	0.9	22
165	Cis-eQTL analysis and functional validation of candidate susceptibility genes for high-grade serous ovarian cancer. Nature Communications, 2015, 6, 8234.	5.8	63
166	Prevalence of PALB2 mutations in Australian familial breast cancer cases and controls. Breast Cancer Research, 2015, 17, 111.	2.2	36
167	A comprehensive evaluation of myocardial fibrosis in hypertrophic cardiomyopathy with cardiac magnetic resonance imaging: linking genotype with fibrotic phenotype. European Heart Journal Cardiovascular Imaging, 2014, 15, 1108-1116.	0.5	77
168	Mutations in RAB39B Cause X-Linked Intellectual Disability and Early-Onset Parkinson Disease with Î±-Synuclein Pathology. American Journal of Human Genetics, 2014, 95, 729-735.	2.6	207
169	Breast-Cancer Risk in Families With Mutations in PALB2. Obstetrical and Gynecological Survey, 2014, 69, 659-660.	0.2	1
170	Associations of common breast cancer susceptibility alleles with risk of breast cancer subtypes in BRCA1 and BRCA2 mutation carriers. Breast Cancer Research, 2014, 16, 3416.	2.2	57
171	Breast-Cancer Risk in Families with Mutations in PALB2. New England Journal of Medicine, 2014, 371, 497-506.	13.9	745
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