Paul James

List of Publications by Year
in descending order

212 papers	10,055 citations	44069 48 h-index	46799 89 g-index
229 all docs	229 docs citations	229 times ranked	15923 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 Study of Sudden Cardiac Death among Children and Young Adults. New England Journal of Medicine, 2016, 374, 2441-2452.	27.0	619
3	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	21.4	356
4	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
5	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	21.4	289
6	Cancer Risks Associated With Germline PALB2 Pathogenic Variants: An International Study of 524 Families. Journal of Clinical Oncology, 2020, 38, 674-685.	1.6	270
7	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581.	21.4	265
8	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
9	Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620.	2.5	224
10	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.	6.2	207
11	Cancer Risks forMLH1andMSH2Mutation Carriers. Human Mutation, 2013, 34, 490-497.	2.5	201
12	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
13	Monogenic and polygenic determinants of sarcoma risk: an international genetic study. Lancet Oncology, The, 2016, 17, 1261-1271.	10.7	161
14	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
15	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.	1.6	152
16	Interim Results from the IMPACT Study: Evidence for Prostate-specific Antigen Screening in BRCA2 Mutation Carriers. European Urology, 2019, 76, 831-842.	1.9	148
17	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
18	Further molecular and clinical delineation of co-locating 17p13.3 microdeletions and microduplications that show distinctive phenotypes. Journal of Medical Genetics, 2010, 47, 299-311.	3.2	137

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19	Mutational Signature Analysis Reveals NTHL1 Deficiency to Cause a Multi-tumor Phenotype. Cancer Cell, 2019, 35, 256-266.e5.	16.8	123
20	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	21.4	120
21	Optimal Selection of Individuals for BRCA Mutation Testing: A Comparison of Available Methods. Journal of Clinical Oncology, 2006, 24, 707-715.	1.6	112
22	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
23	Genome-wide association study identifies a common variant in RAD51B associated with male breast cancer risk. Nature Genetics, 2012, 44, 1182-1184.	21.4	99
24	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
25	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
26	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	12.8	93
27	Real-world utility of whole exome sequencing with targeted gene analysis for focal epilepsy. Epilepsy Research, 2017, 131, 1-8.	1.6	93
28	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
29	The Landscape of Somatic Genetic Alterations in Breast Cancers From ATM Germline Mutation Carriers. Journal of the National Cancer Institute, 2018, 110, 1030-1034.	6.3	90
30	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	12.8	90
31	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.	5.0	88
32	Shared heritability and functional enrichment across six solid cancers. Nature Communications, 2019, 10, 431.	12.8	88
33	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
34	Specifications of the ACMG/AMP variant interpretation guidelines for germline TP53 variants. Human Mutation, 2021, 42, 223-236.	2.5	81
35	Cpipe: a shared variant detection pipeline designed for diagnostic settings. Genome Medicine, 2015, 7, 68.	8.2	78
36	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.	1.2	77

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37	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
38	A Role for Common Genomic Variants in the Assessment of Familial Breast Cancer. Journal of Clinical Oncology, 2012, 30, 4330-4336.	1.6	74
39	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
40	Cis-eQTL analysis and functional validation of candidate susceptibility genes for high-grade serous ovarian cancer. Nature Communications, 2015, 6, 8234.	12.8	63
41	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
42	Copy-number variation associated with congenital anomalies of the kidney and urinary tract. Pediatric Nephrology, 2015, 30, 487-495.	1.7	61
43	The genomic landscape of phaeochromocytoma. Journal of Pathology, 2015, 236, 78-89.	4.5	61
44	Phenotypic variability of distal 22q11.2 copy number abnormalities. American Journal of Medical Genetics, Part A, 2011, 155, 1623-1633.	1.2	59
45	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
46	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.	5.0	57
47	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.	0.9	54
48	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
49	Cardiac arrest and sudden cardiac death registries: a systematic review of global coverage. Open Heart, 2020, 7, e001195.	2.3	52
50	High Frequency of Germline TP53 Mutations in a Prospective Adult-Onset Sarcoma Cohort. PLoS ONE, 2013, 8, e69026.	2.5	51
51	Rare variants in XRCC2 as breast cancer susceptibility alleles: TableÂ1. Journal of Medical Genetics, 2012, 49, 618-620.	3.2	49
52	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
53	Characterization of the Cancer Spectrum in Men With GermlineBRCA1andBRCA2Pathogenic Variants. JAMA Oncology, 2020, 6, 1218.	7.1	48
54	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.	10.7	48

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55	The Medical Genome Reference Bank contains whole genome and phenotype data of 2570 healthy elderly. Nature Communications, 2020, 11, 435.	12.8	47
56	Molecular analysis of PALB2â€associated breast cancers. Journal of Pathology, 2018, 245, 53-60.	4.5	46
57	Population-based genetic testing of asymptomatic women for breast and ovarian cancer susceptibility. Genetics in Medicine, 2019, 21, 913-922.	2.4	45
58	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. Genetics in Medicine, 2022, 24, 130-145.	2.4	45
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.	0.4	42
60	The molecular genetics of non-ALS motor neuron diseases. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2006, 1762, 986-1000.	3.8	41
61	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
62	Cell-type-specific enrichment of risk-associated regulatory elements at ovarian cancer susceptibility loci. Human Molecular Genetics, 2015, 24, 3595-3607.	2.9	40
63	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.9	39
64	Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. American Journal of Human Genetics, 2020, 107, 837-848.	6.2	39
65	Complete overlap of PHACE syndrome and sternal malformation?vascular dysplasia association. American Journal of Medical Genetics Part A, 2002, 110, 78-84.	2.4	37
66	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
67	Prevalence of PALB2 mutations in Australian familial breast cancer cases and controls. Breast Cancer Research, 2015, 17, 111.	5.0	36
68	Diagnostic and cost utility of whole exome sequencing in peripheral neuropathy. Annals of Clinical and Translational Neurology, 2017, 4, 318-325.	3.7	36
69	Current review ofTP53pathogenic germline variants in breast cancer patients outside Li-Fraumeni syndrome. Human Mutation, 2018, 39, 1764-1773.	2.5	35
70	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.	2.4	35
71	Brugada Syndrome Caused by a Large Deletion in SCN5A Only Detected by Multiplex Ligation-Dependent Probe Amplification. Journal of Cardiovascular Electrophysiology, 2011, 22, 1073-1076.	1.7	34
72	The spectrum of BRCA1 and BRCA2 pathogenic sequence variants in Middle Eastern, North African, and South European countries. Human Mutation, 2019, 40, e1-e23.	2.5	34

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73	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.	2.4	34
74	Insights into sudden cardiac death: exploring the potential relevance of non-diagnostic autopsy findings. European Heart Journal, 2019, 40, 831-838.	2.2	33
75	Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€receptor status. Genetic Epidemiology, 2020, 44, 442-468.	1.3	32
76	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.	1.6	30
77	Pheo-Type: A Diagnostic Gene-expression Assay for the Classification of Pheochromocytoma and Paraganglioma. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 1034-1043.	3.6	29
78	Improved, ACMG-compliant, in silico prediction of pathogenicity for missense substitutions encoded by TP53 variants. Human Mutation, 2018, 39, 1061-1069.	2.5	29
79	Crystal structure of human wildtype and S581L-mutant glycyl-tRNA synthetase, an enzyme underlying distal spinal muscular atrophy. FEBS Letters, 2007, 581, 2959-2964.	2.8	28
80	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
81	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38.	5.2	28
82	Prospective Evaluation of the Utility of Whole Exome Sequencing in Dilated Cardiomyopathy. Journal of the American Heart Association, 2020, 9, e013346.	3.7	28
83	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
84	The Angelina Jolie effect. Medical Journal of Australia, 2013, 199, 646-646.	1.7	26
85	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
86	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
87	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
88	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
89	Exome sequencing of familial high-grade serous ovarian carcinoma reveals heterogeneity for rare candidate susceptibility genes. Nature Communications, 2020, 11, 1640.	12.8	24
90	Polygenic risk modeling for prediction of epithelial ovarian cancer risk. European Journal of Human Genetics, 2022, 30, 349-362.	2.8	23

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91	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
92	Prospective validation of the NCI Breast Cancer Risk Assessment Tool (Gail Model) on 40,000 Australian women. Breast Cancer Research, 2018, 20, 155.	5.0	22
93	The consequences of risk reducing salpingo-oophorectomy: the case for a coordinated approach to long-term follow up post surgical menopause. Familial Cancer, 2012, 11, 403-410.	1.9	21
94	Genetic resilience to Alzheimer's disease in APOE Îµ4 homozygotes: A systematic review. Alzheimer's and Dementia, 2019, 15, 1612-1623.	0.8	21
95	A quantitative model to predict pathogenicity of missense variants in the TP53 gene. Human Mutation, 2019, 40, 788-800.	2.5	21
96	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
97	CHARGE association and secondary hypoadrenalism. , 2002, 117A, 177-180.		20
98	Medically actionable pathogenic variants in a population of 13,131 healthy elderly individuals. Genetics in Medicine, 2020, 22, 1883-1886.	2.4	20
99	Tumour morphology predicts PALB2 germline mutation status. British Journal of Cancer, 2013, 109, 154-163.	6.4	19
100	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
101	Mutations in RECQL are not associated with breast cancer risk in an Australian population. Nature Genetics, 2018, 50, 1346-1348.	21.4	19
102	Uptake of polygenic risk information among women at increased risk of breast cancer. Clinical Genetics, 2020, 97, 492-501.	2.0	19
103	Cost-effectiveness of long-term clinical management of BRCA pathogenic variant carriers. Genetics in Medicine, 2020, 22, 831-839.	2.4	19
104	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
105	Cancer Diagnoses Following Abnormal Noninvasive Prenatal Testing: A Case Series, Literature Review, and Proposed Management Model. JCO Precision Oncology, 2021, 5, 1001-1012.	3.0	19
106	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
107	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.	2.5	18
108	The End Unexplained Cardiac Death (EndUCD) Registry for Young Australian Sudden Cardiac Arrest. Heart Lung and Circulation, 2021, 30, 714-720.	0.4	18

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109	Familial Cerebro-costo-mandibular syndrome: a case with unusual prenatal findings and review. Clinical Dysmorphology, 2003, 12, 63-68.	0.3	17
110	Clinical implications of genomics for cancer risk genetics. Lancet Oncology, The, 2015, 16, e303-e308.	10.7	17
111	The Cardiac Genetics Clinic: a model for multidisciplinary genomic medicine. Medical Journal of Australia, 2015, 203, 261-261.	1.7	16
112	Reevaluation of RINT1 as a breast cancer predisposition gene. Breast Cancer Research and Treatment, 2016, 159, 385-392.	2.5	16
113	Relevance of DNA damage repair in the management of prostate cancer. Current Problems in Cancer, 2017, 41, 287-301.	2.0	16
114	High-risk womenâ€™s risk perception after receiving personalized polygenic breast cancer risk information. Journal of Community Genetics, 2019, 10, 197-206.	1.2	16
115	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
116	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
117	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
118	Molecular diagnosis in a pregnancy at risk for both spondyloepiphyseal dysplasia congenita and achondroplasia. Prenatal Diagnosis, 2003, 23, 861-863.	2.3	15
119	Molecular comparison of interval and screenâ€detected breast cancers. Journal of Pathology, 2019, 248, 243-252.	4.5	15
120	Disjunctive Globalization in the Era of the Great Unsettling. Theory, Culture and Society, 2020, 37, 187-203.	2.4	15
121	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.	1.4	15
122	Womenâ€™s responses and understanding of polygenic breast cancer risk information. Familial Cancer, 2020, 19, 297-306.	1.9	15
123	Surveillance in Germline TP53 Mutation Carriers Utilizing Whole-Body Magnetic Resonance Imaging. JAMA Oncology, 2017, 3, 1735.	7.1	14
124	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
125	A synonymous GATA2 variant underlying familial myeloid malignancy with striking intrafamilial phenotypic variability. British Journal of Haematology, 2020, 190, e297-e301.	2.5	14
126	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.	2.4	14

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127	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
128	Another case of interstitial del(12) involving the proposed cardio-facio-cutaneous candidate region. American Journal of Medical Genetics, Part A, 2005, 136A, 12-16.	1.2	13
129	Highâ€level 46XX/46XY chimerism without clinical effect in a healthy multiparous female. American Journal of Medical Genetics, Part A, 2011, 155, 2484-2488.	1.2	13
130	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
131	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
132	The economic impact of sudden cardiac arrest. Resuscitation, 2021, 163, 49-56.	3.0	13
133	Arrhythmic Phenotypes Are a Defining Feature of Dilated Cardiomyopathy-Associated SCN5A Variants: A Systematic Review. Circulation Genomic and Precision Medicine, 2022, 15, CIRCGEN121003432.	3.6	13
134	Prostate-specific antigen velocity in a prospective prostate cancer screening study of men with genetic predisposition. British Journal of Cancer, 2018, 118, 266-276.	6.4	12
135	Common Susceptibility Loci for Male Breast Cancer. Journal of the National Cancer Institute, 2021, 113, 453-461.	6.3	12
136	Communicating polygenic risk scores in the familial breast cancer clinic. Patient Education and Counseling, 2021, 104, 2512-2521.	2.2	12
137	Investigation of monogenic causes of familial breast cancer: data from the BEACCON case-control study. Npj Breast Cancer, 2021, 7, 76.	5.2	12
138	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
139	A severe case of oculo-ectodermal syndrome?. Clinical Dysmorphology, 2002, 11, 179-182.	0.3	11
140	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
141	p53 major hotspot variants are associated with poorer prognostic features in hereditary cancer patients. Cancer Genetics, 2019, 235-236, 21-27.	0.4	11
142	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
143	Prenatal diagnosis of mosaic trisomy 20 in New Zealand. Australian and New Zealand Journal of Obstetrics and Gynaecology, 2002, 42, 486-489.	1.0	10
144	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.	2.6	10

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145	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.	4.1	10
146	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
147	Population DNA screening for medically actionable disease risk in adults. Medical Journal of Australia, 2022, 216, 278-280.	1.7	10
148	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
149	Cousins not twins: intratumoural and intertumoural heterogeneity in syndromic neuroendocrine tumours. Journal of Pathology, 2017, 242, 273-283.	4.5	9
150	Genetic testing in dementiaâ€A medical genetics perspective. International Journal of Geriatric Psychiatry, 2021, 36, 1158-1170.	2.7	9
151	Mendelian randomisation study of smoking exposure in relation to breast cancer risk. British Journal of Cancer, 2021, 125, 1135-1145.	6.4	9
152	Familial mitral valve prolapse associated with short stature, characteristic face, and sudden death. American Journal of Medical Genetics Part A, 2003, 119A, 32-36.	2.4	8
153	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
154	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
155	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
156	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.	0.4	8
157	Familial Aortopathies â€“ State of the Art Review. Heart Lung and Circulation, 2020, 29, 607-618.	0.4	8
158	Predictors and outcomes of in-hospital referrals for forensic investigation after young sudden cardiac death. Heart Rhythm, 2022, 19, 937-944.	0.7	8
159	Spot Diagnosis. New England Journal of Medicine, 2014, 370, 2229-2236.	27.0	7
160	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
161	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
162	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

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163	Partial tetrasomy 15 due to a unique inverted triplication of chromosome15q24-q26. American Journal of Medical Genetics Part A, 2004, 130A, 208-210.	2.4	6
164	Exploring the feasibility and utility of exomeâ€scale tumour sequencing in a clinical setting. Internal Medicine Journal, 2018, 48, 786-794.	0.8	6
165	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
166	Changing landscape of hereditary breast and ovarian cancer germline genetic testing in Australia. Internal Medicine Journal, 2018, 48, 1269-1272.	0.8	6
167	Genomic Risk Prediction for Breast Cancer in Older Women. Cancers, 2021, 13, 3533.	3.7	6
168	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.	2.5	6
169	Estimating single nucleotide polymorphism associations using pedigree data: applications to breast cancer. British Journal of Cancer, 2013, 108, 2610-2622.	6.4	5
170	1p13.2 deletion displays clinical features overlapping Noonan syndrome, likely related to NRAS gene haploinsufficiency. Genetics and Molecular Biology, 2016, 39, 349-357.	1.3	5
171	Cancer risk management in Tasmanian women with BRCA1 and BRCA2 mutations. Familial Cancer, 2018, 17, 333-344.	1.9	5
172	Arrhythmogenic Right Ventricular Cardiomyopathy: A Review of Living and Deceased Probands. Heart Lung and Circulation, 2019, 28, 1034-1041.	0.4	5
173	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
174	Association of germline variation with the survival of women with BRCA1/2 pathogenic variants and breast cancer. Npj Breast Cancer, 2020, 6, 44.	5.2	5
175	Severe musculoskeletal phenotype associated with an unbalanced t(6;10) translocation: Clarification of the locus for this phenotype on distal 6p. American Journal of Medical Genetics Part A, 2003, 119A, 288-292.	2.4	4
176	Breast Conservation Versus Mastectomy in Triple-Negative Breast Cancer: Two Steps Forward, One Step Back?. Journal of Clinical Oncology, 2011, 29, 4722-4723.	1.6	4
177	Metaphors and why these are important in all aspects of genetic counseling. Journal of Genetic Counseling, 2022, 31, 34-40.	1.6	4
178	Polygenic risk in familial breast cancer: Changing the dynamics of communicating genetic risk. Journal of Genetic Counseling, 2022, 31, 120-129.	1.6	4
179	Suspected clonal hematopoiesis as a natural functional assay of TP53 germline variant pathogenicity. Genetics in Medicine, 2022, 24, 673-680.	2.4	4
180	Confirming a diagnosis of hereditary colorectal cancer: the impact of a Familial Bowel Cancer Registry in New Zealand. New Zealand Medical Journal, 2006, 119, U2168.	0.5	4

#	Article	IF	Citations
181	Real world outcomes and implementation pathways of exome sequencing in an adult genetic department. Genetics in Medicine, 2022, , .	2.4	4
182	Population-basedBRCA1/2testing programmes are highly acceptable in the Jewish community: results of the JeneScreen Study. Journal of Medical Genetics, 2023, 60, 265-273.	3.2	4
183	Breast cancer risk is not increased in individuals with TWIST1 mutation confirmed Saethreâ€“Chotzen syndrome: An Australian multicenter study. Genes Chromosomes and Cancer, 2009, 48, 533-538.	2.8	3
184	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.	1.6	3
185	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.	2.2	3
186	Implementing gene curation for hereditary cancer susceptibility in Australia: achieving consensus on genes with clinical utility. Journal of Medical Genetics, 2021, 58, 853-858.	3.2	3
187	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.	2.3	3
188	Globalization in question: why does engaged theory matter?. Globalizations, 2021, 18, 794-809.	2.7	3
189	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
190	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.	1.6	3
191	Interdisciplinary Education for Genetic Counselors: Developing the Concept and Assessing the Need in Australasia. Journal of Genetic Counseling, 2014, 23, 708-724.	1.6	2
192	Population Genomic Screening of All Young Adults in a Health-Care System: A Cost-Effectiveness Analysis. Obstetrical and Gynecological Survey, 2020, 75, 91-93.	0.4	2
193	Can post-mortem coronary artery calcium scores aid diagnosis in young sudden death?. Forensic Science, Medicine, and Pathology, 2021, 17, 27-35.	1.4	2
194	Evaluation of two population screening programmes for BRCA1/2 founder mutations in the Australian Jewish community: a protocol paper. BMJ Open, 2021, 11, e041186.	1.9	2
195	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.	3.2	2
196	Stakeholder attitudes towards establishing a national genomics registry of inherited cancer predisposition: a qualitative study. Journal of Community Genetics, 2022, 13, 59-73.	1.2	2
197	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
198	Towards more effective and equitable genetic testing for BRCA1 and BRCA2 mutaion carriers. Journal of Medical Genetics, 2008, 45, 765-766.	3.2	1

#	Article	IF	Citations
199	Breast-Cancer Risk in Families With Mutations in PALB2. Obstetrical and Gynecological Survey, 2014, 69, 659-660.	0.4	1
200	Heterogeneity and Uncertainties Specific to Genome-Based Health Technological Assessments. Value in Health, 2018, 21, 891-893.	0.3	1
201	Higher risk of phaeochromocytoma/paraganglioma (Phaeoâ€Pgl) in SDHD than SDHB carriers: an Australian cohort study. Internal Medicine Journal, 2019, 49, 529-532.	0.8	1
202	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.	0.4	1
203	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
204	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
205	Contribution of large genomic rearrangements inPALB2to familial breast cancer: implications for genetic testing. Journal of Medical Genetics, 2023, 60, 112-118.	3.2	1
206	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
207	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, , .	1.6	1
208	A comparison of Australian and French families affected by sarcoma: perceptions of genetics and incidental findings. Personalized Medicine, 2018, 15, 13-24.	1.5	0
209	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
210	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
211	Pregnancy Outcomes in Females With Dilated Cardiomyopathyâ€“Associated Rare Genetic Variants. Circulation Genomic and Precision Medicine, 2022, , CIRCGEN121003540.	3.6	0
212	Higher rates but similar causes of young outâ€ofâ€hospital cardiac arrest in rural Australian patients. Australian Journal of Rural Health, 0, , .	1.5	0