Ingrid M Winship

List of Publications by Year
in descending order

237 papers	9,712 citations	53794 45 h-index	45317 90 g-index
251 all docs	251 docs citations	251 times ranked	12296 citing authors

#	Article	IF	Citations
1	Informed choice and attitudes regarding a genomic test to predict risk of colorectal cancer in general practice. Patient Education and Counseling, 2022, 105, 987-995.	2.2	7
2	A step forward, but still inadequate: Australian health professionalsâ€™ views on the genetics and life insurance moratorium. Journal of Medical Genetics, 2022, 59, 817-826.	3.2	6
3	A novel candidate gene in autosomal dominant facial pruritus. Clinical and Experimental Dermatology, 2022, 47, 184-186.	1.3	2
4	Aspirin and the Risk of Colorectal Cancer According to Genetic Susceptibility among Older Individuals. Cancer Prevention Research, 2022, 15, 447-454.	1.5	5
5	Population DNA screening for medically actionable disease risk in adults. Medical Journal of Australia, 2022, 216, 278-280.	1.7	10
6	Population-based estimates of age-specific cumulative risk of breast cancer for pathogenic variants in ATM. Breast Cancer Research, 2022, 24, 24.	5.0	3
7	Weight is More Informative than Body Mass Index for Predicting Postmenopausal Breast Cancer Risk: Prospective Family Study Cohort (ProF-SC). Cancer Prevention Research, 2022, 15, 185-191.	1.5	4
8	Real world outcomes and implementation pathways of exome sequencing in an adult genetic department. Genetics in Medicine, 2022, , .	2.4	4
9	Germline MBD4 deficiency causes a multi-tumor predisposition syndrome. American Journal of Human Genetics, 2022, 109, 953-960.	6.2	23
10	Rivaroxaban in the treatment of <scp>TEK</scp> â€related venous malformation. Australasian Journal of Dermatology, 2022, , .	0.7	2
11	Identifying colorectal cancer caused by biallelic MUTYH pathogenic variants using tumor mutational signatures. Nature Communications, 2022, 13, .	12.8	15
12	Cancer risk and tumour spectrum in 172 patients with a germlineSUFUpathogenic variation: a collaborative study of the SIOPE Host Genome Working Group. Journal of Medical Genetics, 2022, 59, 1123-1132.	3.2	4
13	Heterogeneity in the psychosocial and behavioral responses associated with a diagnosis of suspected Lynch syndrome in women with endometrial cancer. Hereditary Cancer in Clinical Practice, 2022, 20, .	1.5	3
14	Analysis in the Prospective Lynch Syndrome Database identifies sarcoma as part of the Lynch syndrome tumor spectrum. International Journal of Cancer, 2021, 148, 512-513.	5.1	9
15	Risk-reducing hysterectomy and bilateral salpingo-oophorectomy in female heterozygotes of pathogenic mismatch repair variants: a Prospective Lynch Syndrome Database report. Genetics in Medicine, 2021, 23, 705-712.	2.4	28
16	Integrated Guidance for Enhancing the Care of Familial Hypercholesterolaemia in Australia. Heart Lung and Circulation, 2021, 30, 324-349.	0.4	51
17	Polygenic score modifies risk for Alzheimer's disease in APOE Î¼4 homozygotes at phenotypic extremes. Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring, 2021, 13, e12226.	2.4	7
18	Evaluating the utility of tumour mutational signatures for identifying hereditary colorectal cancer and polyposis syndrome carriers. Gut, 2021, 70, 2138-2149.	12.1	27

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19	The Management of Peutzâ€“Jeghers Syndrome: European Hereditary Tumour Group (EHTG) Guideline. Journal of Clinical Medicine, 2021, 10, 473.	2.4	65
20	Mainstreaming genomics: training experience of hospital medical officers at the Royal Melbourne Hospital. Internal Medicine Journal, 2021, 51, 268-271.	0.8	2
21	Genetic testing in dementiaâ€A medical genetics perspective. International Journal of Geriatric Psychiatry, 2021, 36, 1158-1170.	2.7	9
22	Prospective Evaluation of the Addition of Polygenic Risk Scores to Breast Cancer Risk Models. JNCI Cancer Spectrum, 2021, 5, pkab021.	2.9	19
23	Germline and Tumor Sequencing as a Diagnostic Tool To Resolve Suspected Lynch Syndrome. Journal of Molecular Diagnostics, 2021, 23, 358-371.	2.8	12
24	Population-Based Estimates of the Age-Specific Cumulative Risk of Breast Cancer for Pathogenic Variants in CHEK2: Findings from the Australian Breast Cancer Family Registry. Cancers, 2021, 13, 1378.	3.7	5
25	Assessment of a Polygenic Risk Score for Colorectal Cancer to Predict Risk of Lynch Syndrome Colorectal Cancer. JNCI Cancer Spectrum, 2021, 5, pkab022.	2.9	15
26	Uptake of hysterectomy and bilateral salpingo-oophorectomy in carriers of pathogenic mismatch repair variants: a Prospective Lynch Syndrome Database report. European Journal of Cancer, 2021, 148, 124-133.	2.8	11
27	Study protocol: the Australian genetics and life insurance moratoriumâ€"monitoring the effectiveness and response (A-GLIMMER) project. BMC Medical Ethics, 2021, 22, 63.	2.4	12
28	Population-based estimates of the age-specific cumulative risk of breast cancer for pathogenic variants in CHEK2: Findings from the Australian Breast Cancer Family Registry.. Journal of Clinical Oncology, 2021, 39, 551-551.	1.6	1
29	DNA Methylation Signatures and the Contribution of Age-Associated Methylomic Drift to Carcinogenesis in Early-Onset Colorectal Cancer. Cancers, 2021, 13, 2589.	3.7	18
30	Multifocal extracardiac rhabdomyomas: extending the phenotype of Birtâ€“Hoggâ€“DubÃ© syndrome. British Journal of Dermatology, 2021, 185, 861-863.	1.5	3
31	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
32	No Difference in Penetrance between Truncating and Missense/Aberrant Splicing Pathogenic Variants in MLH1 and MSH2: A Prospective Lynch Syndrome Database Study. Journal of Clinical Medicine, 2021, 10, 2856.	2.4	11
33	Genomic Risk Prediction for Breast Cancer in Older Women. Cancers, 2021, 13, 3533.	3.7	6
34	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
35	Translation of a circulating miRNA signature of melanoma into a solid tissue assay to improve diagnostic accuracy and precision. Biomarkers in Medicine, 2021, 15, 1111-1122.	1.4	4
36	Genetic variant interpretation: a primer for clinicians. Internal Medicine Journal, 2021, 51, 1401-1406.	0.8	0

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37	Intent vs. impact: calling time on outdated phenotypic labels in dermatology. British Journal of Dermatology, 2021, , .	1.5	0
38	â€œLeft in limboâ€: Exploring how patients with colorectal cancer interpret and respond to a suspected Lynch syndrome diagnosis. Hereditary Cancer in Clinical Practice, 2021, 19, 43.	1.5	3
39	Prospective Evaluation over 15 Years of Six Breast Cancer Risk Models. Cancers, 2021, 13, 5194.	3.7	7
40	Rare germline variants in the AXIN2 gene in families with colonic polyposis and colorectal cancer. Familial Cancer, 2021, , 1.	1.9	5
41	Making community voices heard in a researchâ€“health service alliance, the evolving role of the Community Advisory Group: a case study from the membersâ€™ perspective. Research Involvement and Engagement, 2021, 7, 84.	2.9	3
42	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
43	Cancer risks by gene, age, and gender in 6350 carriers of pathogenic mismatch repair variants: findings from the Prospective Lynch Syndrome Database. Genetics in Medicine, 2020, 22, 15-25.	2.4	365
44	Genetic mosaicism in dermatology: Clinical utility of genetic testing of skin lesions. Australasian Journal of Dermatology, 2020, 61, 92-94.	0.7	1
45	More than meets the eye: Palmoplantar keratoderma and arrhythmogenic right ventricular cardiomyopathy in a patient with loss of the DSP gene. JAAD Case Reports, 2020, 6, 804-806.	0.8	2
46	Second International Guidelines for the Diagnosis and Management of Hereditary Hemorrhagic Telangiectasia. Annals of Internal Medicine, 2020, 173, 989-1001.	3.9	244
47	A Genomic Test for Colorectal Cancer Risk: Is This Acceptable and Feasible in Primary Care?. Public Health Genomics, 2020, 23, 110-121.	1.0	12
48	Risk-Reducing Gynecological Surgery in Lynch Syndrome: Results of an International Survey from the Prospective Lynch Syndrome Database. Journal of Clinical Medicine, 2020, 9, 2290.	2.4	12
49	Is CHEK2 a moderate-risk breast cancer gene or the younger sister of Li-Fraumeni?. BMJ Case Reports, 2020, 13, e236435.	0.5	5
50	The Impact of a Comprehensive Risk Prediction Model for Colorectal Cancer on a Population Screening Program. JNCI Cancer Spectrum, 2020, 4, pkaa062.	2.9	5
51	Phenotypic discordance between siblings with junctional epidermolysis bullosaâ€“pyloric atresia. Clinical and Experimental Dermatology, 2020, 45, 793-795.	1.3	2
52	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
53	Medically actionable pathogenic variants in a population of 13,131 healthy elderly individuals. Genetics in Medicine, 2020, 22, 1883-1886.	2.4	20
54	Mismatch repair gene pathogenic germline variants in a population-based cohort of breast cancer. Familial Cancer, 2020, 19, 197-202.	1.9	6

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55	Persistent Troponin Elevation in Left-Dominant Arrhythmogenic Cardiomyopathy. Circulation Genomic and Precision Medicine, 2020, 13, e003094.	3.6	4
56	Arrhythmogenic Right Ventricular Cardiomyopathy: A Review of Living and Deceased Probands. Heart Lung and Circulation, 2019, 28, 1034-1041.	0.4	5
57	Response to Veenstra et al.. Genetics in Medicine, 2019, 21, 2842-2843.	2.4	6
58	The Use of Optimal Treatment for DLBCL Is Improving in All Age Groups and Is a Key Factor in Overall Survival, but Non-Clinical Factors Influence Treatment. Cancers, 2019, 11, 928.	3.7	5
59	Survival by colon cancer stage and screening interval in Lynch syndrome: a prospective Lynch syndrome database report. Hereditary Cancer in Clinical Practice, 2019, 17, 28.	1.5	27
60	Genetic resilience to Alzheimer's disease in APOE Îµ4 homozygotes: A systematic review. Alzheimer's and Dementia, 2019, 15, 1612-1623.	0.8	21
61	Type 2 diabetes mellitus, blood cholesterol, triglyceride and colorectal cancer risk in Lynch syndrome. British Journal of Cancer, 2019, 121, 869-876.	6.4	10
62	Phenotypic confirmation of oligodontia, colorectal polyposis and cancer in a family carrying an exon 7 nonsense variant in the AXIN2 gene. Familial Cancer, 2019, 18, 311-315.	1.9	20
63	Loss-of-Function in SMAD4 Might Not Be Critical for Human Natural Killer Cell Responsiveness to TGF-Î². Frontiers in Immunology, 2019, 10, 904.	4.8	0
64	Living with Hereditary Haemorrhagic Telangiectasia: stigma, coping with unpredictable symptoms, and self-advocacy. Psychology and Health, 2019, 34, 1141-1160.	2.2	6
65	An alternative approach to establishing unbiased colorectal cancer risk estimation in Lynch syndrome. Genetics in Medicine, 2019, 21, 2706-2712.	2.4	11
66	Ability of known susceptibility SNPs to predict colorectal cancer risk for persons with and without a family history. Familial Cancer, 2019, 18, 389-397.	1.9	23
67	Tumor mutational signatures in sebaceous skin lesions from individuals with Lynch syndrome. Molecular Genetics & Genomic Medicine, 2019, 7, e00781.	1.2	8
68	Incident Chronic Kidney Disease After Radical Nephrectomy for Renal Cell Carcinoma. Clinical Genitourinary Cancer, 2019, 17, e581-e591.	1.9	3
69	Warfarin ineffective as symptomatic therapy for erythropoietic protoporphyria. Australasian Journal of Dermatology, 2019, 60, e360-e361.	0.7	2
70	Lack of association between screening interval and cancer stage in Lynch syndrome may be accounted for by over-diagnosis; a prospective Lynch syndrome database report. Hereditary Cancer in Clinical Practice, 2019, 17, 8.	1.5	42
71	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
72	Phacomatosis pigmentokeratotica: Postzygotic <scp>HRAS</scp>Âmutation with malignant degeneration of the sebaceous naevus. Australasian Journal of Dermatology, 2019, 60, e245-e246.	0.7	5

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73	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
74	Healthcare System-Funded Preventive Genomic Screening: Challenges for Australia and Other Single-Payer Systems. Public Health Genomics, 2019, 22, 140-144.	1.0	6
75	The influence of obesity-related factors in the etiology of renal cell carcinomaâ€"A mendelian randomization study. PLoS Medicine, 2019, 16, e1002724.	8.4	59
76	Insights into sudden cardiac death: exploring the potential relevance of non-diagnostic autopsy findings. European Heart Journal, 2019, 40, 831-838.	2.2	33
77	Chondrodysplasia punctata (<scp>CDPX</scp>2) in a male caused by singleâ€gene mosaicism: A 20â€year followâ€up. Australasian Journal of Dermatology, 2019, 60, e160-e162.	0.7	3
78	Clinicoâ€pathological predictors of mismatch repair deficiency in sebaceous neoplasia: A large case series from a single Australian private pathology service. Australasian Journal of Dermatology, 2019, 60, 126-133.	0.7	13
79	Assessing the ProMCol classifier as a prognostic marker for non-metastatic colorectal cancer within the Melbourne Collaborative Cohort Study. Gut, 2019, 68, 761-762.	12.1	2
80	Hemangioblastoma in Hereditary Leiomyomatosis and Renal Cell Cancer Syndrome: a phenotypic overlap between VHL and HLRCC Syndromes. Familial Cancer, 2019, 18, 91-95.	1.9	1
81	Update on Hereditary Colorectal Cancer: Improving the Clinical Utility of Multigene Panel Testing. Clinical Colorectal Cancer, 2018, 17, e293-e305.	2.3	55
82	Mutations in SUFU and PTCH1 genes may cause different cutaneous cancer predisposition syndromes: similar, but not the same. Familial Cancer, 2018, 17, 601-606.	1.9	23
83	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
84	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
85	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
86	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
87	Genomic Characterization of Upper-Tract Urothelial Carcinoma in Patients With Lynch Syndrome. JCO Precision Oncology, 2018, 2018, 1-13.	3.0	29
88	Cancer Risks for PMS2-Associated Lynch Syndrome. Journal of Clinical Oncology, 2018, 36, 2961-2968.	1.6	147
89	Absence of renal phenotype in hereditary haemorrhagic telangiectasia. Internal Medicine Journal, 2018, 48, 1255-1257.	0.8	3
90	Maximizing the Clinical Benefit of DPYD Genotyping: Extending the Opportunity of Personalized Management to Family Members Through Cascade Testing. JCO Precision Oncology, 2018, 2, 1-5.	3.0	0

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91	The use of a risk assessment and decision support tool (CRISP) compared with usual care in general practice to increase risk-stratified colorectal cancer screening: study protocol for a randomised controlled trial. Trials, 2018, 19, 397.	1.6	13
92	Utility of immunohistochemistry for mismatch repair proteins on colorectal polyps in the familial cancer clinic. Internal Medicine Journal, 2018, 48, 1325-1330.	0.8	2
93	Family historyâ€"based colorectal cancer screening in Australia: A modelling study of the costs, benefits, and harms of different participation scenarios. PLoS Medicine, 2018, 15, e1002630.	8.4	6
94	APRT deficiency: the need for early diagnosis. BMJ Case Reports, 2018, 2018, bcr-2018-225742.	0.5	3
95	The role of <scp>STK 11</scp> gene testing in individuals with oral pigmentation. Australasian Journal of Dermatology, 2017, 58, 135-138.	0.7	12
96	Tumor testing to identify lynch syndrome in two Australian colorectal cancer cohorts. Journal of Gastroenterology and Hepatology (Australia), 2017, 32, 427-438.	2.8	47
97	Mutation screening of ACKR3 and COPS8 in kidney cancer cases from the CONFIRM study. Familial Cancer, 2017, 16, 411-416.	1.9	6
98	The course of anxiety, depression and unmet needs in survivors of diffuse large B cell lymphoma and multiple myeloma in the early survivorship period. Journal of Cancer Survivorship, 2017, 11, 329-338.	2.9	25
99	Multiple cutaneous leiomyomas leading to discovery of novel splice mutation in the fumarate hydratase gene associated with <scp>HLRCC</scp>. Australasian Journal of Dermatology, 2017, 58, e246-e248.	0.7	3
100	Preparing for genomic medicine: a real world demonstration of health system change. Npj Genomic Medicine, 2017, 2, 16.	3.8	73
101	Trends in the surgical management of stage 1 renal cell carcinoma: findings from a populationâ€based study. BJU International, 2017, 120, 6-14.	2.5	19
102	Genome-wide association study identifies multiple risk loci for renal cell carcinoma. Nature Communications, 2017, 8, 15724.	12.8	106
103	Cohort study of Gorlin syndrome with emphasis on standardised phenotyping and quality of life assessment. Internal Medicine Journal, 2017, 47, 664-673.	0.8	14
104	Pathogenic variants in the healthy elderly: unique ethical and practical challenges. Journal of Medical Ethics, 2017, 43, 714-722.	1.8	10
105	Lack of evidence for germline RNF43 mutations in patients with serrated polyposis syndrome from a large multinational study. Gut, 2017, 66, 1170-1172.	12.1	42
106	Distress and unmet needs during treatment and quality of life in early cancer survivorship: A longitudinal study of haematological cancer patients. European Journal of Haematology, 2017, 99, 423-430.	2.2	43
107	Penetrance and the Healthy Elderly. Genetic Testing and Molecular Biomarkers, 2017, 21, 637-640.	0.7	3
108	Rare disease registries: a call to action. Internal Medicine Journal, 2017, 47, 1075-1079.	0.8	33

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109	The influence of unmet supportive care needs on anxiety and depression during cancer treatment and beyond: a longitudinal study of survivors of haematological cancers. Supportive Care in Cancer, 2017, 25, 3447-3456.	2.2	33
110	Meanings of abortion in context: accounts of abortion in the lives of women diagnosed with breast cancer. BMC Women's Health, 2017, 17, 26.	2.0	6
111	Alcohol Consumption and the Risk of Colorectal Cancer for Mismatch Repair Gene Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2017, 26, 366-375.	2.5	37
112	Consent Processes for Mobile App Mediated Research: Systematic Review. JMIR MHealth and UHealth, 2017, 5, e126.	3.7	33
113	Gene panel testing for hereditary breast cancer. Medical Journal of Australia, 2016, 204, 188-190.	1.7	12
114	Risk factors for metachronous colorectal cancer following a primary colorectal cancer: A prospective cohort study. International Journal of Cancer, 2016, 139, 1081-1090.	5.1	32
115	Multivitamin, calcium and folic acid supplements and the risk of colorectal cancer in Lynch syndrome. International Journal of Epidemiology, 2016, 45, 940-953.	1.9	27
116	Renal angiomyolipoma in Birtâ€“Hoggâ€“Dube syndrome: A case study supporting overlap with tuberous sclerosis complex. American Journal of Medical Genetics, Part A, 2016, 170, 3323-3326.	1.2	8
117	Caregiversâ€™ information needs and their â€˜experiences of careâ€™ during treatment are associated with elevated anxiety and depression: a cross-sectional study of the caregivers of renal cancer survivors. Supportive Care in Cancer, 2016, 24, 4177-4186.	2.2	34
118	A Prospective Study of Sudden Cardiac Death among Children and Young Adults. New England Journal of Medicine, 2016, 374, 2441-2452.	27.0	619
119	PALB2: research reaching to clinical outcomes for women with breast cancer. Hereditary Cancer in Clinical Practice, 2016, 14, 9.	1.5	27
120	SNP rs16906252C>T Is an Expression and Methylation Quantitative Trait Locus Associated with an Increased Risk of Developing MGMT-Methylated Colorectal Cancer. Clinical Cancer Research, 2016, 22, 6266-6277.	7.0	22
121	Risk of extracolonic cancers for people with biallelic and monoallelic mutations in MUTYH. International Journal of Cancer, 2016, 139, 1557-1563.	5.1	107
122	Mal de <scp>M</scp>eleda in <scp>I</scp>ndonesia: Mutations in the <scp>SLURP</scp>1 gene appear to be ubiquitous. Australasian Journal of Dermatology, 2016, 57, e11-3.	0.7	8
123	Multiple familial pilomatrixomas in the absence of other clinical features: a case of familial benign pilomatrixoma. Australasian Journal of Dermatology, 2016, 57, 75-76.	0.7	3
124	Germline mutations inPMS2andMLH1in individuals with solitary loss of PMS2 expression in colorectal carcinomas from the Colon Cancer Family Registry Cohort. BMJ Open, 2016, 6, e010293.	1.9	33
125	Incidental diagnosis of HLRCC following investigation for Asperger Syndrome: actionable and actioned. Familial Cancer, 2016, 15, 25-29.	1.9	4
126	Outcomes of a randomised controlled trial of a complex genetic counselling intervention to improve family communication. European Journal of Human Genetics, 2016, 24, 356-360.	2.8	55

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127	Red cells from ferrochelatase-deficient erythropoietic protoporphyria patients are resistant to growth of malarial parasites. Blood, 2015, 125, 534-541.	1.4	37
128	Halo naevi and cafÃ© au lait macule regression in a renal transplant patient on immunosuppression. Australasian Journal of Dermatology, 2015, 56, e88-90.	0.7	0
129	Fertility after youngâ€onset colorectal cancer: a study of subjects with Lynch syndrome. Colorectal Disease, 2015, 17, 787-793.	1.4	17
130	Lynch syndrome and cervical cancer. International Journal of Cancer, 2015, 137, 2757-2761.	5.1	13
131	Precision medicine: are we there?. Medical Journal of Australia, 2015, 203, 132-133.	1.7	3
132	The Cardiac Genetics Clinic: a model for multidisciplinary genomic medicine. Medical Journal of Australia, 2015, 203, 261-261.	1.7	16
133	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
134	Is cardiac rhabdomyoma a feature of Birt Hogg DubÃ© syndrome?. American Journal of Medical Genetics, Part A, 2015, 167, 802-804.	1.2	7
135	Sudden unexpected death, epilepsy and familial cardiac pathology. Journal of Clinical Neuroscience, 2015, 22, 1594-1600.	1.5	9
136	Risk of colorectal cancer for people with a mutation in both a MUTYH and a DNA mismatch repair gene. Familial Cancer, 2015, 14, 575-583.	1.9	11
137	Aspirin, Ibuprofen, and the Risk of Colorectal Cancer in Lynch Syndrome. Journal of the National Cancer Institute, 2015, 107, djv170.	6.3	80
138	Female Hormonal Factors and the Risk of Endometrial Cancer in Lynch Syndrome. JAMA - Journal of the American Medical Association, 2015, 314, 61.	7.4	68
139	Multiple skin hamartomata: a possible novel clinical presentation of SUFU neoplasia syndrome. Familial Cancer, 2015, 14, 151-155.	1.9	15
140	Implementing a telephone based peer support intervention for women with a BRCA1/2 mutation. Familial Cancer, 2015, 14, 373-382.	1.9	7
141	Interpretation of genomic variation and disease association: the great missense mutation challenge!. Breast Cancer Research and Treatment, 2015, 151, 475-476.	2.5	0
142	Vestibular schwannoma in a patient with neurofibromatosis type 1: clinical report and literature review. Familial Cancer, 2015, 14, 157-160.	1.9	2
143	Genetic variants within the hTERT gene and the risk of colorectal cancer in Lynch syndrome. Genes and Cancer, 2015, 6, 445-451.	1.9	6
144	Rare genetic variants: making the connection with breast cancer susceptibility. AIMS Genetics, 2015, 02, 281-292.	1.9	2

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145	Randomized Controlled Trial of a Telephone-Based Peer-Support Program for Women Carrying aBRCA1orBRCA2Mutation: Impact on Psychological Distress. Journal of Clinical Oncology, 2014, 32, 4073-4080.	1.6	24
146	High prevalence of mismatch repair deficiency in prostate cancers diagnosed in mismatch repair gene mutation carriers from the colon cancer family registry. Familial Cancer, 2014, 13, 573-582.	1.9	44
147	Re: Microsatellite Instability and BRAF Mutation Testing in Colorectal Cancer Prognostication. Journal of the National Cancer Institute, 2014, 106, dju180-dju180.	6.3	6
148	Appreciating the broad clinical features of SMAD4 mutation carriers: a multicenter chart review. Genetics in Medicine, 2014, 16, 588-593.	2.4	62
149	Dermatitis Artefacta Presenting as a Recurrent Skin Eruption in a Patient with 1p36 Deletion Syndrome. Australasian Journal of Dermatology, 2014, 55, 90-90.	0.7	1
150	The future in clinical genetics: affective forecasting biases in patient and clinician decision making. Clinical Genetics, 2014, 85, 312-317.	2.0	33
151	Improving family communication after a new genetic diagnosis: a randomised controlled trial of a genetic counselling intervention. BMC Medical Genetics, 2014, 15, 33.	2.1	26
152	Succinate Dehydrogenase Deficiency Is Rare in Pituitary Adenomas. American Journal of Surgical Pathology, 2014, 38, 560-566.	3.7	71
153	Hereditary haemorrhagic telangiectasia, an <scp>A</scp>ustralian cohort: clinical and investigative features. Internal Medicine Journal, 2014, 44, 639-644.	0.8	12
154	Risk of Colorectal Cancer for Carriers of Mutations in MUTYH, WithÂand Without a Family History of Cancer. Gastroenterology, 2014, 146, 1208-1211.e5.	1.3	180
155	Should the grading of colorectal adenocarcinoma include microsatellite instability status?. Human Pathology, 2014, 45, 2077-2084.	2.0	44
156	Fertility and apparent genetic anticipation in Lynch syndrome. Familial Cancer, 2014, 13, 369-374.	1.9	3
157	Prevalence of PALB2 mutations in Australasian multiple-case breast cancer families. Breast Cancer Research, 2013, 15, R17.	5.0	42
158	Clinical predictors of genetic testing outcomes in hypertrophic cardiomyopathy. Genetics in Medicine, 2013, 15, 972-977.	2.4	110
159	Are the common genetic variants associated with colorectal cancer risk for DNA mismatch repair gene mutation carriers?. European Journal of Cancer, 2013, 49, 1578-1587.	2.8	31
160	Risks of Colorectal and Other Cancers After Endometrial Cancer for Women With Lynch Syndrome. Journal of the National Cancer Institute, 2013, 105, 274-279.	6.3	93
161	Coâ€existence of hereditary coproporphyria and porphyria cutanea tarda: The importance of genetic testing. Australasian Journal of Dermatology, 2013, 54, e50-2.	0.7	1
162	FSH receptor gene variants are rarely associated with premature ovarian failure. Reproductive BioMedicine Online, 2013, 26, 396-399.	2.4	22

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163	Familial cardiological and targeted genetic evaluation: Low yield in sudden unexplained death and high yield in unexplained cardiac arrest syndromes. Heart Rhythm, 2013, 10, 1653-1660.	0.7	83
164	Risk of Metachronous Colon Cancer Following Surgery for Rectal Cancer in Mismatch Repair Gene Mutation Carriers. Annals of Surgical Oncology, 2013, 20, 1829-1836.	1.5	103
165	Deficiency in Complement Factor B. New England Journal of Medicine, 2013, 369, 1667-1669.	27.0	52
166	Unmet support needs and distress among women with a BRCA1/2 mutation. Familial Cancer, 2013, 12, 509-518.	1.9	21
167	Family History of Colorectal Cancer in BRAF p.V600E-Mutated Colorectal Cancer Cases. Cancer Epidemiology Biomarkers and Prevention, 2013, 22, 917-926.	2.5	24
168	FamilialSDHAMutation Associated With Pituitary Adenoma and Pheochromocytoma/Paraganglioma. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E1103-E1108.	3.6	102
169	Systematic Review of Quality Improvement Interventions Directed at Cancer Specialists. Journal of Clinical Oncology, 2013, 31, 1583-1591.	1.6	12
170	Fertility Management After Breast Cancer Diagnosis: A Qualitative Investigation of Women's Experiences of and Recommendations for Professional Care. Health Care for Women International, 2013, 34, 50-67.	1.1	26
171	The utility of genetics in inherited cancer. Medical Journal of Australia, 2013, 199, 644-644.	1.7	1
172	A funding model for publicâ€good clinical trials. Medical Journal of Australia, 2013, 199, 90-91.	1.7	2
173	PALB2 and breast cancer: ready for clinical translation!. The Application of Clinical Genetics, 2013, 6, 43.	3.0	40
174	Screening Practices of Unaffected People at Familial Risk of Colorectal Cancer. Cancer Prevention Research, 2012, 5, 240-247.	1.5	25
175	Colorectal and Other Cancer Risks for Carriers and Noncarriers From Families With a DNA Mismatch Repair Gene Mutation: A Prospective Cohort Study. Journal of Clinical Oncology, 2012, 30, 958-964.	1.6	286
176	Risks of Primary Extracolonic Cancers Following Colorectal Cancer in Lynch Syndrome. Journal of the National Cancer Institute, 2012, 104, 1363-1372.	6.3	193
177	Phenotype in novel Xp duplication. American Journal of Medical Genetics, Part A, 2012, 158A, 2342-2346.	1.2	6
178	Screening practices of Australian men and women categorized as â€œat or slightly above average riskâ€• of colorectal cancer. Cancer Causes and Control, 2012, 23, 1853-1864.	1.8	17
179	Colorectal Cancer Linkage on Chromosomes 4q21, 8q13, 12q24, and 15q22. PLoS ONE, 2012, 7, e38175.	2.5	24
180	The Australasian Colorectal Cancer Family Registry. Medical Journal of Australia, 2012, 197, 480-481.	1.7	13

#	Article	IF	Citations
181	Xâ€linked recessive polyfibromatosis manifesting with spontaneous keloid scars and Dupuytren's contracture. Australasian Journal of Dermatology, 2012, 53, 148-150.	0.7	6
182	Metachronous colorectal cancer risk for mismatch repair gene mutation carriers: the advantage of more extensive colon surgery. Gut, 2011, 60, 950-957.	12.1	227
183	Renal Tumors and Hereditary Pheochromocytoma-Paraganglioma Syndrome Type 4. New England Journal of Medicine, 2011, 364, 885-886.	27.0	120
184	Melanoma(s) arising in large segmental speckled lentiginous nevi: AÂcase series. Journal of the American Academy of Dermatology, 2011, 64, 1190-1193.	1.2	12
185	Renal Tumors Associated With Germline SDHB Mutation Show Distinctive Morphology. American Journal of Surgical Pathology, 2011, 35, 1578-1585.	3.7	184
186	Immunohistochemistry to Detect Hereditary Nonpolyposis Colorectal Cancer in Young Patients: the 7-Year Auckland Experience. Diseases of the Colon and Rectum, 2011, 54, 552-558.	1.3	16
187	A photographic essay of prolidase deficiency. Clinical Dysmorphology, 2011, 20, 194-199.	0.3	13
188	Body Mass Index in Early Adulthood and Endometrial Cancer Risk for Mismatch Repair Gene Mutation Carriers. Obstetrics and Gynecology, 2011, 117, 899-905.	2.4	23
189	Practical guidelines addressing ethical issues pertaining to the curation of human locus-specific variation databases (LSDBs). Human Mutation, 2010, 31, 1179-1184.	2.5	36
190	Risks of Lynch Syndrome Cancers for MSH6 Mutation Carriers. Journal of the National Cancer Institute, 2010, 102, 193-201.	6.3	328
191	A PALB2 mutation associated with high risk of breast cancer. Breast Cancer Research, 2010, 12, R109.	5.0	102
192	Antioxidant Effect of Warfarin Therapy: A Possible Symptomatic Treatment for Erythropoietic Protoporphyria. Archives of Dermatology, 2009, 145, 960-1.	1.4	5
193	Cultural enhancement of a clinical service to meet the needs of indigenous people; genetic service development in response to issues for New Zealand Maori. Clinical Genetics, 2008, 73, 132-138.	2.0	25
194	Familial breast cancer: double heterozygosity for BRCA1 and BRCA2 mutations with differing phenotypes. Familial Cancer, 2008, 7, 119-124.	1.9	21
195	The Clinical Phenotype of Lynch Syndrome Due to Germ-Line PMS2 Mutations. Gastroenterology, 2008, 135, 419-428.e1.	1.3	480
196	Lessons from the skinâ€”cutaneous features of familial cancer. Lancet Oncology, The, 2008, 9, 462-472.	10.7	43
197	Towards more effective and equitable genetic testing for BRCA1 and BRCA2 mutation carriers. Journal of Medical Genetics, 2008, 45, 409-410.	3.2	1
198	Suggested actions from the Melbourne HVP Information Seminar. Nature Precedings, 2008, , .	0.1	0

#	Article	IF	Citations
199	Suggested actions from the Melbourne HVP Information Seminar. Nature Precedings, 2008, , .	0.1	0
200	Polymorphism in intron 1 of the interferon-gamma gene influences both serum immunoglobulin E levels and the risk for chronic hepatitis B virus infection in Polynesians. Immunogenetics, 2007, 59, 187-195.	2.4	24
201	Hereditary Diffuse Gastric Cancer: Diagnosis and Management. Clinical Gastroenterology and Hepatology, 2006, 4, 262-275.	4.4	163
202	Hyperplastic Polyposis Syndrome: Phenotypic Presentations and the Role of MBD4 and MYH. Gastroenterology, 2006, 131, 30-39.	1.3	186
203	Mutational analysis of BMP15 and GDF9 as candidate genes for premature ovarian failure. Fertility and Sterility, 2006, 86, 1009-1012.	1.0	75
204	Mutational screening of FOXO3A and FOXO1A in women with premature ovarian failure. Fertility and Sterility, 2006, 86, 1518-1521.	1.0	106
205	Genetic Diversity and Linkage Disequilibrium in the Polynesian Population of Niue Island. Human Biology, 2006, 78, 131-145.	0.2	7
206	Autosomal recessive Alport syndrome: an in-depth clinical and molecular analysis of five families. Nephrology Dialysis Transplantation, 2006, 21, 665-671.	0.7	40
207	An investigation into FOXE1 polyalanine tract length in premature ovarian failure. Molecular Human Reproduction, 2006, 12, 145-149.	2.8	17
208	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
209	Blepharocheilodontic syndrome or lagophthalmos: a child with overlapping features. Clinical Dysmorphology, 2005, 14, 151-153.	0.3	0
210	Further case report of a child with a 9q34 deletion and a review of the reported cases. American Journal of Medical Genetics, Part A, 2005, 133A, 219-221.	1.2	3
211	INHA promoter polymorphisms are associated with premature ovarian failure. Molecular Human Reproduction, 2005, 11, 779-784.	2.8	55
212	Controversies and ethical issues in cancer-genetics clinics. Lancet Oncology, The, 2005, 6, 301-310.	10.7	67
213	Blepharocheilodontic syndrome or lagophthalmos: a child with overlapping features. Clinical Dysmorphology, 2005, 14, 151-153.	0.3	0
214	Asthma phenotypes in Niue Islanders. Respirology, 2004, 9, 521-527.	2.3	1
215	False-positive diagnosis of trisomy 21 using fluorescencein situ hybridisation (FISH) on uncultured amniotic fluid cells. Prenatal Diagnosis, 2003, 23, 302-305.	2.3	7
216	Identification of novel mutations in FOXL2 associated with premature ovarian failure. Molecular Human Reproduction, 2002, 8, 729-733.	2.8	154

#	Article	IF	Citations
217	Supernumerary marker chromosomes 5: Confirmation of a critical region and resultant phenotype. American Journal of Medical Genetics Part A, 2002, 111, 19-26.	2.4	25
218	Novel germlineCDH1mutations in hereditary diffuse gastric cancer families. Human Mutation, 2002, 19, 518-525.	2.5	63
219	Analysis of the TGF Î² functional pathway in epithelial ovarian carcinoma. British Journal of Cancer, 2001, 85, 687-691.	6.4	28
220	Clinical and Molecular Genetic Features of Pulmonary Hypertension in Patients with Hereditary Hemorrhagic Telangiectasia. New England Journal of Medicine, 2001, 345, 325-334.	27.0	676
221	Hierarchical mutation screening protocol for theBRCA1 gene. Human Mutation, 2000, 16, 422-430.	2.5	2
222	Null Alleles at the Huntington Disease Locus: Implications for Diagnostics and CAG Repeat Instability. Genetic Testing and Molecular Biomarkers, 2000, 4, 55-60.	1.7	18
223	Inhibin: a candidate gene for premature ovarian failure. Human Reproduction, 2000, 15, 2644-2649.	0.9	168
224	DNA microsatellite instability and mismatch repair protein loss in adenomas presenting in hereditary non-polyposis colorectal cancer. Gut, 2000, 47, 37-42.	12.1	149
225	X-Linked Late-Onset Sensorineural Deafness Caused by a Deletion Involving OA1 and a Novel Gene Containing WD-40 Repeats. American Journal of Human Genetics, 1999, 64, 1604-1616.	6.2	97
226	Mixed epithelial polyps in association with hereditary non-polyposis colorectal cancer providing an alternative pathway of cancer histogenesis. Pathology, 1997, 29, 28-33.	0.6	77
227	X-Linked Ocular Albinism and Sensorineural Deafness: Linkage to Xp22.3. Genomics, 1993, 18, 444-445.	2.9	26
228	Hearing Impairment and Pigmentary Disturbance. Annals of the New York Academy of Sciences, 1991, 630, 152-166.	3.8	26
229	Primary oxalosis-an unusual cause of livedo reticularis. Clinical and Experimental Dermatology, 1991, 16, 367-370.	1.3	17
230	Microcephaly-cardiomyopathy: a new autosomal recessive phenotype?. Journal of Medical Genetics, 1991, 28, 619-621.	3.2	5
231	Piebaldism: an autonomous autosomal dominant entity. Clinical Genetics, 1991, 39, 330-337.	2.0	20
232	Genetic heterogeneity in tuberous sclerosis: phenotypic correlations.. Journal of Medical Genetics, 1990, 27, 418-421.	3.2	13
233	Epidermolysis bullosa misdiagnosed as child abuse. A report of 3 cases. South African Medical Journal, 1988, 73, 369-70.	0.6	5
234	Sotos syndrome â€" autosomal dominant inheritance substantiated. Clinical Genetics, 1985, 28, 243-246.	2.0	43

#	Article	IF	Citations
235	X-linked inheritance of ocular albinism with late-onset sensorineural deafness. American Journal of Medical Genetics Part A, 1984, 19, 797-803.	2.4	21
236	Suggested actions from the Melbourne HVP Information Seminar. Nature Precedings, 0, , .	0.1	1
237	The role of cutaneous manifestations in the diagnosis of the Ehlersâ€Danlos syndromes. Skin Health and Disease, 0, , .	1.5	1