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	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
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	The Clinical Phenotype of Lynch Syndrome Due to Germ-Line PMS2 Mutations. Gastroenterology, 2008, 135, 419-428.e1.	1.3	480
4	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
5	Risks of Lynch Syndrome Cancers for MSH6 Mutation Carriers. Journal of the National Cancer Institute, 2010, 102, 193-201.	6.3	328
6	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
7	Second International Guidelines for the Diagnosis and Management of Hereditary Hemorrhagic Telangiectasia. Annals of Internal Medicine, 2020, 173, 989-1001.	3.9	244
8	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
9	Risks of Primary Extracolonic Cancers Following Colorectal Cancer in Lynch Syndrome. Journal of the National Cancer Institute, 2012, 104, 1363-1372.	6.3	193
10	Hyperplastic Polyposis Syndrome: Phenotypic Presentations and the Role of MBD4 and MYH. Gastroenterology, 2006, 131, 30-39.	1.3	186
11	Renal Tumors Associated With Germline SDHB Mutation Show Distinctive Morphology. American Journal of Surgical Pathology, 2011, 35, 1578-1585.	3.7	184
12	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
13	Inhibin: a candidate gene for premature ovarian failure. Human Reproduction, 2000, 15, 2644-2649.	0.9	168
14	Hereditary Diffuse Gastric Cancer: Diagnosis and Management. Clinical Gastroenterology and Hepatology, 2006, 4, 262-275.	4.4	163
15	Identification of novel mutations in FOXL2 associated with premature ovarian failure. Molecular Human Reproduction, 2002, 8, 729-733.	2.8	154
16	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
17	Cancer Risks for PMS2-Associated Lynch Syndrome. Journal of Clinical Oncology, 2018, 36, 2961-2968.	1.6	147
18	Renal Tumors and Hereditary Pheochromocytoma-Paraganglioma Syndrome Type 4. New England Journal of Medicine, 2011, 364, 885-886.	27.0	120

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19	Clinical predictors of genetic testing outcomes in hypertrophic cardiomyopathy. Genetics in Medicine, 2013, 15, 972-977.	2.4	110
20	Risk of extracolonic cancers for people with biallelic and monoallelic mutations in MUTYH. International Journal of Cancer, 2016, 139, 1557-1563.	5.1	107
21	Mutational screening of FOXO3A and FOXO1A in women with premature ovarian failure. Fertility and Sterility, 2006, 86, 1518-1521.	1.0	106
22	Genome-wide association study identifies multiple risk loci for renal cell carcinoma. Nature Communications, 2017, 8, 15724.	12.8	106
23	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
24	A PALB2 mutation associated with high risk of breast cancer. Breast Cancer Research, 2010, 12, R109.	5.0	102
25	FamilialSDHAMutation Associated With Pituitary Adenoma and Pheochromocytoma/Paraganglioma. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E1103-E1108.	3.6	102
26	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
27	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
28	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
29	Aspirin, Ibuprofen, and the Risk of Colorectal Cancer in Lynch Syndrome. Journal of the National Cancer Institute, 2015, 107, djv170.	6.3	80
30	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
31	Mutational analysis of BMP15 and GDF9 as candidate genes for premature ovarian failure. Fertility and Sterility, 2006, 86, 1009-1012.	1.0	75
32	Preparing for genomic medicine: a real world demonstration of health system change. Npj Genomic Medicine, 2017, 2, 16.	3.8	73
33	Succinate Dehydrogenase Deficiency Is Rare in Pituitary Adenomas. American Journal of Surgical Pathology, 2014, 38, 560-566.	3.7	71
34	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
35	Controversies and ethical issues in cancer-genetics clinics. Lancet Oncology, The, 2005, 6, 301-310.	10.7	67
36	The Management of Peutzâ€"Jeghers Syndrome: European Hereditary Tumour Group (EHTG) Guideline. Journal of Clinical Medicine, 2021, 10, 473.	2.4	65

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37	Novel germlineCDH1mutations in hereditary diffuse gastric cancer families. Human Mutation, 2002, 19, 518-525.	2.5	63
38	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
39	Appreciating the broad clinical features of SMAD4 mutation carriers: a multicenter chart review. Genetics in Medicine, 2014, 16, 588-593.	2.4	62
40	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
41	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
42	INHA promoter polymorphisms are associated with premature ovarian failure. Molecular Human Reproduction, 2005, 11, 779-784.	2.8	55
43	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
44	Update on Hereditary Colorectal Cancer: Improving the Clinical Utility of Multigene Panel Testing. Clinical Colorectal Cancer, 2018, 17, e293-e305.	2.3	55
45	Deficiency in Complement Factor B. New England Journal of Medicine, 2013, 369, 1667-1669.	27.0	52
46	Integrated Guidance for Enhancing the Care of Familial Hypercholesterolaemia in Australia. Heart Lung and Circulation, 2021, 30, 324-349.	0.4	51
47	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
48	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
49	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
50	Should the grading of colorectal adenocarcinoma include microsatellite instability status?. Human Pathology, 2014, 45, 2077-2084.	2.0	44
51	Sotos syndrome â€" autosomal dominant inheritance substantiated. Clinical Genetics, 1985, 28, 243-246.	2.0	43
52	Lessons from the skinâ€"cutaneous features of familial cancer. Lancet Oncology, The, 2008, 9, 462-472.	10.7	43
53	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
54	Prevalence of PALB2 mutations in Australasian multiple-case breast cancer families. Breast Cancer Research, 2013, 15, R17.	5.0	42

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55	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
56	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
57	Autosomal recessive Alport syndrome: an in-depth clinical and molecular analysis of five families. Nephrology Dialysis Transplantation, 2006, 21, 665-671.	0.7	40
58	PALB2 and breast cancer: ready for clinical translation!. The Application of Clinical Genetics, 2013, 6, 43.	3.0	40
59	Red cells from ferrochelatase-deficient erythropoietic protoporphyria patients are resistant to growth of malarial parasites. Blood, 2015, 125, 534-541.	1.4	37
60	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
61	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
62	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
63	The future in clinical genetics: affective forecasting biases in patient and clinician decision making. Clinical Genetics, 2014, 85, 312-317.	2.0	33
64	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
65	Rare disease registries: a call to action. Internal Medicine Journal, 2017, 47, 1075-1079.	0.8	33
66	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
67	Insights into sudden cardiac death: exploring the potential relevance of non-diagnostic autopsy findings. European Heart Journal, 2019, 40, 831-838.	2.2	33
68	Consent Processes for Mobile App Mediated Research: Systematic Review. JMIR MHealth and UHealth, 2017, 5, e126.	3.7	33
69	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
70	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
71	Genomic Characterization of Upper-Tract Urothelial Carcinoma in Patients With Lynch Syndrome. JCO Precision Oncology, 2018, 2018, 1-13.	3.0	29
72	Analysis of the TGF Î² functional pathway in epithelial ovarian carcinoma. British Journal of Cancer, 2001, 85, 687-691.	6.4	28

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73	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
74	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
75	PALB2: research reaching to clinical outcomes for women with breast cancer. Hereditary Cancer in Clinical Practice, 2016, 14, 9.	1.5	27
76	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
77	Evaluating the utility of tumour mutational signatures for identifying hereditary colorectal cancer and polyposis syndrome carriers. Gut, 2021, 70, 2138-2149.	12.1	27
78	Hearing Impairment and Pigmentary Disturbance. Annals of the New York Academy of Sciences, 1991, 630, 152-166.	3.8	26
79	X-Linked Ocular Albinism and Sensorineural Deafness: Linkage to Xp22.3. Genomics, 1993, 18, 444-445.	2.9	26
80	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
81	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
82	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
83	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
84	Screening Practices of Unaffected People at Familial Risk of Colorectal Cancer. Cancer Prevention Research, 2012, 5, 240-247.	1.5	25
85	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
86	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
87	Colorectal Cancer Linkage on Chromosomes 4q21, 8q13, 12q24, and 15q22. PLoS ONE, 2012, 7, e38175.	2.5	24
88	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
89	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
90	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

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91	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
92	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
93	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
94	Germline MBD4 deficiency causes a multi-tumor predisposition syndrome. American Journal of Human Genetics, 2022, 109, 953-960.	6.2	23
95	FSH receptor gene variants are rarely associated with premature ovarian failure. Reproductive BioMedicine Online, 2013, 26, 396-399.	2.4	22
96	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
97	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
98	Familial breast cancer: double heterozygosity for BRCA1 and BRCA2 mutations with differing phenotypes. Familial Cancer, 2008, 7, 119-124.	1.9	21
99	Unmet support needs and distress among women with a BRCA1/2 mutation. Familial Cancer, 2013, 12, 509-518.	1.9	21
100	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
101	Genetic resilience to Alzheimer's disease in APOE Îµ4 homozygotes: A systematic review. Alzheimer's and Dementia, 2019, 15, 1612-1623.	0.8	21
102	Piebaldism: an autonomous autosomal dominant entity. Clinical Genetics, 1991, 39, 330-337.	2.0	20
103	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
104	Medically actionable pathogenic variants in a population of 13,131 healthy elderly individuals. Genetics in Medicine, 2020, 22, 1883-1886.	2.4	20
105	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
106	Prospective Evaluation of the Addition of Polygenic Risk Scores to Breast Cancer Risk Models. JNCI Cancer Spectrum, 2021, 5, pkab021.	2.9	19
107	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
108	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

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109	Primary oxalosis-an unusual cause of livedo reticularis. Clinical and Experimental Dermatology, 1991, 16, 367-370.	1.3	17
110	An investigation into FOXE1 polyalanine tract length in premature ovarian failure. Molecular Human Reproduction, 2006, 12, 145-149.	2.8	17
111	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
112	Fertility after youngâ€•onset colorectal cancer: a study of subjects with Lynch syndrome. Colorectal Disease, 2015, 17, 787-793.	1.4	17
113	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
114	The Cardiac Genetics Clinic: a model for multidisciplinary genomic medicine. Medical Journal of Australia, 2015, 203, 261-261.	1.7	16
115	Multiple skin hamartomata: a possible novel clinical presentation of SUFU neoplasia syndrome. Familial Cancer, 2015, 14, 151-155.	1.9	15
116	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
117	Identifying colorectal cancer caused by biallelic MUTYH pathogenic variants using tumor mutational signatures. Nature Communications, 2022, 13, .	12.8	15
118	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
119	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
120	Genetic heterogeneity in tuberous sclerosis: phenotypic correlations.. Journal of Medical Genetics, 1990, 27, 418-421.	3.2	13
121	A photographic essay of prolidase deficiency. Clinical Dysmorphology, 2011, 20, 194-199.	0.3	13
122	The Australasian Colorectal Cancer Family Registry. Medical Journal of Australia, 2012, 197, 480-481.	1.7	13
123	Lynch syndrome and cervical cancer. International Journal of Cancer, 2015, 137, 2757-2761.	5.1	13
124	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
125	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
126	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

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127	Systematic Review of Quality Improvement Interventions Directed at Cancer Specialists. Journal of Clinical Oncology, 2013, 31, 1583-1591.	1.6	12
128	Hereditary haemorrhagic telangiectasia, an <scp>A</scp>ustralian cohort: clinical and investigative features. Internal Medicine Journal, 2014, 44, 639-644.	0.8	12
129	Gene panel testing for hereditary breast cancer. Medical Journal of Australia, 2016, 204, 188-190.	1.7	12
130	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
131	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
132	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
133	Germline and Tumor Sequencing as a Diagnostic Tool To Resolve Suspected Lynch Syndrome. Journal of Molecular Diagnostics, 2021, 23, 358-371.	2.8	12
134	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
135	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
136	An alternative approach to establishing unbiased colorectal cancer risk estimation in Lynch syndrome. Genetics in Medicine, 2019, 21, 2706-2712.	2.4	11
137	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
138	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
139	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
140	Pathogenic variants in the healthy elderly: unique ethical and practical challenges. Journal of Medical Ethics, 2017, 43, 714-722.	1.8	10
141	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
142	Population DNA screening for medically actionable disease risk in adults. Medical Journal of Australia, 2022, 216, 278-280.	1.7	10
143	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
144	Sudden unexpected death, epilepsy and familial cardiac pathology. Journal of Clinical Neuroscience, 2015, 22, 1594-1600.	1.5	9

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145	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
146	Genetic testing in dementiaâ€A medical genetics perspective. International Journal of Geriatric Psychiatry, 2021, 36, 1158-1170.	2.7	9
147	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
148	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
149	Tumor mutational signatures in sebaceous skin lesions from individuals with Lynch syndrome. Molecular Genetics & Genomic Medicine, 2019, 7, e00781.	1.2	8
150	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
151	Genetic Diversity and Linkage Disequilibrium in the Polynesian Population of Niue Island. Human Biology, 2006, 78, 131-145.	0.2	7
152	Is cardiac rhabdomyoma a feature of Birt Hogg DubÃ© syndrome?. American Journal of Medical Genetics, Part A, 2015, 167, 802-804.	1.2	7
153	Implementing a telephone based peer support intervention for women with a BRCA1/2 mutation. Familial Cancer, 2015, 14, 373-382.	1.9	7
154	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
155	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
156	Prospective Evaluation over 15 Years of Six Breast Cancer Risk Models. Cancers, 2021, 13, 5194.	3.7	7
157	Phenotype in novel Xp duplication. American Journal of Medical Genetics, Part A, 2012, 158A, 2342-2346.	1.2	6
158	Xâ€linked recessive polyfibromatosis manifesting with spontaneous keloid scars and Dupuytren's contracture. Australasian Journal of Dermatology, 2012, 53, 148-150.	0.7	6
159	Re: Microsatellite Instability and BRAF Mutation Testing in Colorectal Cancer Prognostication. Journal of the National Cancer Institute, 2014, 106, dju180-dju180.	6.3	6
160	Mutation screening of ACKR3 and COPS8 in kidney cancer cases from the CONFIRM study. Familial Cancer, 2017, 16, 411-416.	1.9	6
161	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
162	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

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163	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
164	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
165	Response to Veenstra et al.. Genetics in Medicine, 2019, 21, 2842-2843.	2.4	6
166	Living with Hereditary Haemorrhagic Telangiectasia: stigma, coping with unpredictable symptoms, and self-advocacy. Psychology and Health, 2019, 34, 1141-1160.	2.2	6
167	Healthcare System-Funded Preventive Genomic Screening: Challenges for Australia and Other Single-Payer Systems. Public Health Genomics, 2019, 22, 140-144.	1.0	6
168	Mismatch repair gene pathogenic germline variants in a population-based cohort of breast cancer. Familial Cancer, 2020, 19, 197-202.	1.9	6
169	Genomic Risk Prediction for Breast Cancer in Older Women. Cancers, 2021, 13, 3533.	3.7	6
170	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
171	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
172	Microcephaly-cardiomyopathy: a new autosomal recessive phenotype?. Journal of Medical Genetics, 1991, 28, 619-621.	3.2	5
173	Antioxidant Effect of Warfarin Therapy: A Possible Symptomatic Treatment for Erythropoietic Protoporphyria. Archives of Dermatology, 2009, 145, 960-1.	1.4	5
174	Arrhythmogenic Right Ventricular Cardiomyopathy: A Review of Living and Deceased Probands. Heart Lung and Circulation, 2019, 28, 1034-1041.	0.4	5
175	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
176	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
177	Is CHEK2 a moderate-risk breast cancer gene or the younger sister of Li-Fraumeni?. BMJ Case Reports, 2020, 13, e236435.	0.5	5
178	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
179	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
180	Rare germline variants in the AXIN2 gene in families with colonic polyposis and colorectal cancer. Familial Cancer, 2021, , 1.	1.9	5

#	Article	IF	Citations
181	Epidermolysis bullosa misdiagnosed as child abuse. A report of 3 cases. South African Medical Journal, 1988, 73, 369-70.	0.6	5
182	Aspirin and the Risk of Colorectal Cancer According to Genetic Susceptibility among Older Individuals. Cancer Prevention Research, 2022, 15, 447-454.	1.5	5
183	Incidental diagnosis of HLRCC following investigation for Asperger Syndrome: actionable and actioned. Familial Cancer, 2016, 15, 25-29.	1.9	4
184	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
185	Persistent Troponin Elevation in Left-Dominant Arrhythmogenic Cardiomyopathy. Circulation Genomic and Precision Medicine, 2020, 13, e003094.	3.6	4
186	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
187	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
188	Real world outcomes and implementation pathways of exome sequencing in an adult genetic department. Genetics in Medicine, 2022, , .	2.4	4
189	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
190	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
191	Fertility and apparent genetic anticipation in Lynch syndrome. Familial Cancer, 2014, 13, 369-374.	1.9	3
192	Precision medicine: are we there?. Medical Journal of Australia, 2015, 203, 132-133.	1.7	3
193	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
194	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
195	Penetrance and the Healthy Elderly. Genetic Testing and Molecular Biomarkers, 2017, 21, 637-640.	0.7	3
196	Absence of renal phenotype in hereditary haemorrhagic telangiectasia. Internal Medicine Journal, 2018, 48, 1255-1257.	0.8	3
197	Incident Chronic Kidney Disease After Radical Nephrectomy for Renal Cell Carcinoma. Clinical Genitourinary Cancer, 2019, 17, e581-e591.	1.9	3
198	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

#	Article	IF	Citations
199	Multifocal extracardiac rhabdomyomas: extending the phenotype of Birtâ€“Hoggâ€“DubÃ© syndrome. British Journal of Dermatology, 2021, 185, 861-863.	1.5	3
200	APRT deficiency: the need for early diagnosis. BMJ Case Reports, 2018, 2018, bcr-2018-225742.	0.5	3
201	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
202	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
203	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
204	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
205	Hierarchical mutation screening protocol for theBRCA1 gene. Human Mutation, 2000, 16, 422-430.	2.5	2
206	A funding model for publicâ€good clinical trials. Medical Journal of Australia, 2013, 199, 90-91.	1.7	2
207	Vestibular schwannoma in a patient with neurofibromatosis type 1: clinical report and literature review. Familial Cancer, 2015, 14, 157-160.	1.9	2
208	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
209	Warfarin ineffective as symptomatic therapy for erythropoietic protoporphyria. Australasian Journal of Dermatology, 2019, 60, e360-e361.	0.7	2
210	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
211	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
212	Phenotypic discordance between siblings with junctional epidermolysis bullosaâ€“pyloric atresia. Clinical and Experimental Dermatology, 2020, 45, 793-795.	1.3	2
213	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
214	Mainstreaming genomics: training experience of hospital medical officers at the Royal Melbourne Hospital. Internal Medicine Journal, 2021, 51, 268-271.	0.8	2
215	A novel candidate gene in autosomal dominant facial pruritus. Clinical and Experimental Dermatology, 2022, 47, 184-186.	1.3	2
216	Rare genetic variants: making the connection with breast cancer susceptibility. AIMS Genetics, 2015, 02, 281-292.	1.9	2

#	Article	IF	Citations
217	Rivaroxaban in the treatment of <scp>TEK</scp> â€related venous malformation. Australasian Journal of Dermatology, 2022, , .	0.7	2
218	Asthma phenotypes in Niue Islanders. Respirology, 2004, 9, 521-527.	2.3	1
219	Towards more effective and equitable genetic testing for BRCA1 and BRCA2 mutation carriers. Journal of Medical Genetics, 2008, 45, 409-410.	3.2	1
220	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
221	The utility of genetics in inherited cancer. Medical Journal of Australia, 2013, 199, 644-644.	1.7	1
222	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
223	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
224	Genetic mosaicism in dermatology: Clinical utility of genetic testing of skin lesions. Australasian Journal of Dermatology, 2020, 61, 92-94.	0.7	1
225	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
226	Suggested actions from the Melbourne HVP Information Seminar. Nature Precedings, 0, , .	0.1	1
227	The role of cutaneous manifestations in the diagnosis of the Ehlersâ€Danlos syndromes. Skin Health and Disease, 0, , .	1.5	1
228	Blepharocheilodontic syndrome or lagophthalmos: a child with overlapping features. Clinical Dysmorphology, 2005, 14, 151-153.	0.3	0
229	Suggested actions from the Melbourne HVP Information Seminar. Nature Precedings, 2008, , .	0.1	0
230	Suggested actions from the Melbourne HVP Information Seminar. Nature Precedings, 2008, , .	0.1	0
231	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
232	Interpretation of genomic variation and disease association: the great missense mutation challenge!. Breast Cancer Research and Treatment, 2015, 151, 475-476.	2.5	0
233	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
234	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

#	Article	IF	Citations
235	Genetic variant interpretation: a primer for clinicians. Internal Medicine Journal, 2021, 51, 1401-1406.	0.8	0
236	Intent vs. impact: calling time on outdated phenotypic labels in dermatology. British Journal of Dermatology, 2021, , .	1.5	0
237	Blepharocheilodontic syndrome or lagophthalmos: a child with overlapping features. Clinical Dysmorphology, 2005, 14, 151-153.	0.3	0