Gareth Evans

List of Publications by Citations

955 papers	66,771 citations	120 h-index	230 g-index
1,056 ext. papers	78,022 ext. citations	7.1 avg, IF	7.24 L-index

#	Paper	IF	Citations
955	Average risks of breast and ovarian cancer associated with BRCA1 or BRCA2 mutations detected in case Series unselected for family history: a combined analysis of 22 studies. *American Journal of Human Genetics*, **2003**, 72, 1117-30	11	2643
954	Genome-wide association study identifies novel breast cancer susceptibility loci. *Nature*, **2007**, 447, 1087-93	59.4	1957
953	Replication of genome-wide association signals in UK samples reveals risk loci for type 2 diabetes. *Science*, **2007**, 316, 1336-41	33.3	1823
952	Prophylactic oophorectomy in carriers of BRCA1 or BRCA2 mutations. *New England Journal of Medicine*, **2002**, 346, 1616-22	59.2	1363
951	Risks of Breast, Ovarian, and Contralateral Breast Cancer for BRCA1 and BRCA2 Mutation Carriers. *JAMA - Journal of the American Medical Association*, **2017**, 317, 2402-2416	27.4	1140
950	Association of risk-reducing surgery in BRCA1 or BRCA2 mutation carriers with cancer risk and mortality. *JAMA - Journal of the American Medical Association*, **2010**, 304, 967-75	27.4	993
949	Bilateral prophylactic mastectomy reduces breast cancer risk in BRCA1 and BRCA2 mutation carriers: the PROSE Study Group. *Journal of Clinical Oncology*, **2004**, 22, 1055-62	2.2	924
948	Low-penetrance susceptibility to breast cancer due to CHEK2(*)1100delC in noncarriers of BRCA1 or BRCA2 mutations. *Nature Genetics*, **2002**, 31, 55-9	36.3	863
947	Guidelines for colorectal cancer screening and surveillance in moderate and high risk groups (update from 2002). *Gut*, **2010**, 59, 666-89	19.2	843
946	PALB2, which encodes a BRCA2-interacting protein, is a breast cancer susceptibility gene. *Nature Genetics*, **2007**, 39, 165-7	36.3	719
945	Long-term effect of aspirin on cancer risk in carriers of hereditary colorectal cancer: an analysis from the CAPP2 randomised controlled trial. *Lancet, The*, **2011**, 378, 2081-7	40	715
944	Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. *Nature*, **2010**, 464, 713-20	50.4	639
943	Cancer risks for BRCA1 and BRCA2 mutation carriers: results from prospective analysis of EMBRACE. *Journal of the National Cancer Institute*, **2013**, 105, 812-22	9.7	616
942	Guidelines for the diagnosis and management of individuals with neurofibromatosis 1. *Journal of Medical Genetics*, **2007**, 44, 81-8	5.8	600
941	Gene-panel sequencing and the prediction of breast-cancer risk. *New England Journal of Medicine*, **2015**, 372, 2243-57	59.2	587
940	Genome-wide association study identifies five new breast cancer susceptibility loci. *Nature Genetics* , **2010**, 42, 504-7	36.3	582
939	Identification of the familial cylindromatosis tumour-suppressor gene. *Nature Genetics*, **2000**, 25, 160-5	36.3	560

938	ATM mutations that cause ataxia-telangiectasia are breast cancer susceptibility alleles. *Nature Genetics*, **2006**, 38, 873-5	36.3	553
937	Truncating mutations in the Fanconi anemia J gene BRIP1 are low-penetrance breast cancer susceptibility alleles. *Nature Genetics*, **2006**, 38, 1239-41	36.3	553
936	Oral contraceptives and the risk of hereditary ovarian cancer. Hereditary Ovarian Cancer Clinical Study Group. *New England Journal of Medicine*, **1998**, 339, 424-8	59.2	514
935	Prediction of BRCA1 status in patients with breast cancer using estrogen receptor and basal phenotype. *Clinical Cancer Research*, **2005**, 11, 5175-80	12.9	511
934	Birth incidence and prevalence of tumor-prone syndromes: estimates from a UK family genetic register service. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 327-32	2.5	506
933	Risk-reducing salpingo-oophorectomy for the prevention of BRCA1- and BRCA2-associated breast and gynecologic cancer: a multicenter, prospective study. *Journal of Clinical Oncology*, **2008**, 26, 1331-7	2.2	465
932	A genome-wide association study identifies colorectal cancer susceptibility loci on chromosomes 10p14 and 8q23.3. *Nature Genetics*, **2008**, 40, 623-30	36.3	463
931	Germline BRCA mutations are associated with higher risk of nodal involvement, distant metastasis, and poor survival outcomes in prostate cancer. *Journal of Clinical Oncology*, **2013**, 31, 1748-57	2.2	440
930	Germline mutations of the BRCA1 gene in breast and ovarian cancer families provide evidence for a genotype-phenotype correlation. *Nature Genetics*, **1995**, 11, 428-33	36.3	439
929	Association between BRCA1 and BRCA2 mutations and survival in women with invasive epithelial ovarian cancer. *JAMA - Journal of the American Medical Association*, **2012**, 307, 382-90	27.4	427
928	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. *Nature Genetics*, **2013**, 45, 371-84, 384e1-2	36.3	422
927	The effects of intermittent or continuous energy restriction on weight loss and metabolic disease risk markers: a randomized trial in young overweight women. *International Journal of Obesity*, **2011**, 35, 714-27	5.5	413
926	Pathology of breast and ovarian cancers among BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA). *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 134-47	4	411
925	Newly discovered breast cancer susceptibility loci on 3p24 and 17q23.2. *Nature Genetics*, **2009**, 41, 585-90	36.3	393
924	Complications of the naevoid basal cell carcinoma syndrome: results of a population based study. *Journal of Medical Genetics*, **1993**, 30, 460-4	5.8	389
923	Germline mutations in RAD51D confer susceptibility to ovarian cancer. *Nature Genetics*, **2011**, 43, 879-882	36.3	379
922	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. *American Journal of Human Genetics*, **2019**, 104, 21-34	11	363
921	Relative frequency and morphology of cancers in carriers of germline TP53 mutations. *Oncogene*, **2001**, 20, 4621-8	9.2	361

920	The BOADICEA model of genetic susceptibility to breast and ovarian cancers: updates and extensions. *British Journal of Cancer*, **2008**, 98, 1457-66	8.7	358
919	Autosomal recessive colorectal adenomatous polyposis due to inherited mutations of MYH. *Lancet, The*, **2003**, 362, 39-41	40	358
918	Allele losses in the region 17q12-21 in familial breast and ovarian cancer involve the wild-type chromosome. *Nature Genetics*, **1992**, 2, 128-31	36.3	346
917	Effect of short-term hormone replacement therapy on breast cancer risk reduction after bilateral prophylactic oophorectomy in BRCA1 and BRCA2 mutation carriers: the PROSE Study Group. *Journal of Clinical Oncology*, **2005**, 23, 7804-10	2.2	339
916	Location of gene for Gorlin syndrome. *Lancet, The*, **1992**, 339, 581-2	40	336
915	Assessing women at high risk of breast cancer: a review of risk assessment models. *Journal of the National Cancer Institute*, **2010**, 102, 680-91	9.7	334
914	A clinical study of type 2 neurofibromatosis. *The Quarterly Journal of Medicine*, **1992**, 84, 603-18		323
913	A genetic study of type 2 neurofibromatosis in the United Kingdom. I. Prevalence, mutation rate, fitness, and confirmation of maternal transmission effect on severity. *Journal of Medical Genetics*, **1992**, 29, 841-6	5.8	320
912	Diagnostic criteria for schwannomatosis. *Neurology*, **2005**, 64, 1838-45	6.5	309
911	Meta-analysis of three genome-wide association studies identifies susceptibility loci for colorectal cancer at 1q41, 3q26.2, 12q13.13 and 20q13.33. *Nature Genetics*, **2010**, 42, 973-7	36.3	301
910	Neurofibromatosis type 2 (NF2): a clinical and molecular review. *Orphanet Journal of Rare Diseases*, **2009**, 4, 16	4.2	294
909	Cancer incidence and survival in Lynch syndrome patients receiving colonoscopic and gynaecological surveillance: first report from the prospective Lynch syndrome database. *Gut*, **2017**, 66, 464-472	19.2	291
908	Mortality after bilateral salpingo-oophorectomy in BRCA1 and BRCA2 mutation carriers: a prospective cohort study. *Lancet Oncology, The*, **2006**, 7, 223-9	21.7	291
907	Association of type and location of BRCA1 and BRCA2 mutations with risk of breast and ovarian cancer. *JAMA - Journal of the American Medical Association*, **2015**, 313, 1347-61	27.4	286
906	Tumor risks and genotype-phenotype-proteotype analysis in 358 patients with germline mutations in SDHB and SDHD. *Human Mutation*, **2010**, 31, 41-51	4.7	286
905	An absence of cutaneous neurofibromas associated with a 3-bp inframe deletion in exon 17 of the NF1 gene (c.2970-2972 delAAT): evidence of a clinically significant NF1 genotype-phenotype correlation. *American Journal of Human Genetics*, **2007**, 80, 140-51	11	279
904	A locus on 19p13 modifies risk of breast cancer in BRCA1 mutation carriers and is associated with hormone receptor-negative breast cancer in the general population. *Nature Genetics*, **2010**, 42, 885-92	36.3	276
903	Li-Fraumeni syndrome--a molecular and clinical review. *British Journal of Cancer*, **1997**, 76, 1-14	8.7	274

902	Germline E-cadherin gene (CDH1) mutations predispose to familial gastric cancer and colorectal cancer. *Human Molecular Genetics*, **1999**, 8, 607-10	5.6	271
901	Oral contraceptives and the risk of breast cancer in BRCA1 and BRCA2 mutation carriers. *Journal of the National Cancer Institute*, **2002**, 94, 1773-9	9.7	266
900	Second primary tumors in neurofibromatosis 1 patients treated for optic glioma: substantial risks after radiotherapy. *Journal of Clinical Oncology*, **2006**, 24, 2570-5	2.2	263
899	Cancer risk and survival in carriers by gene and gender up to 75 years of age: a report from the Prospective Lynch Syndrome Database. *Gut*, **2018**, 67, 1306-1316	19.2	259
898	Incidence of vestibular schwannoma and neurofibromatosis 2 in the North West of England over a 10-year period: higher incidence than previously thought. *Otology and Neurotology*, **2005**, 26, 93-7	2.6	255
897	Critical research gaps and translational priorities for the successful prevention and treatment of breast cancer. *Breast Cancer Research*, **2013**, 15, R92	8.3	248
896	Cancer Screening Recommendations for Individuals with Li-Fraumeni Syndrome. *Clinical Cancer Research*, **2017**, 23, e38-e45	12.9	245
895	Psychosocial impact of breast/ovarian (BRCA1/2) cancer-predictive genetic testing in a UK multi-centre clinical cohort. *British Journal of Cancer*, **2004**, 91, 1787-94	8.7	245
894	Cumulative lifetime incidence of extracolonic cancers in Lynch syndrome: a report of 121 families with proven mutations. *Clinical Genetics*, **2009**, 75, 141-9	4	243
893	The incidence of Gorlin syndrome in 173 consecutive cases of medulloblastoma. *British Journal of Cancer*, **1991**, 64, 959-61	8.7	242
892	The effect of intermittent energy and carbohydrate restriction v. daily energy restriction on weight loss and metabolic disease risk markers in overweight women. *British Journal of Nutrition*, **2013**, 110, 1534-47	3.6	229
891	Effect of aspirin or resistant starch on colorectal neoplasia in the Lynch syndrome. *New England Journal of Medicine*, **2008**, 359, 2567-78	59.2	228
890	Common breast cancer-predisposition alleles are associated with breast cancer risk in BRCA1 and BRCA2 mutation carriers. *American Journal of Human Genetics*, **2008**, 82, 937-48	11	218
889	Evaluation of breast cancer risk assessment packages in the family history evaluation and screening programme. *Journal of Medical Genetics*, **2003**, 40, 807-14	5.8	217
888	Germline SDHD mutation in familial phaeochromocytoma. *Lancet, The*, **2001**, 357, 1181-2	40	211
887	Are there low-penetrance TP53 Alleles? evidence from childhood adrenocortical tumors. *American Journal of Human Genetics*, **1999**, 65, 995-1006	11	211
886	Genome-wide association study in BRCA1 mutation carriers identifies novel loci associated with breast and ovarian cancer risk. *PLoS Genetics*, **2013**, 9, e1003212	6	209
885	The Angelina Jolie effect: how high celebrity profile can have a major impact on provision of cancer related services. *Breast Cancer Research*, **2014**, 16, 442	8.3	207

884	Effect of BRCA Mutations on Metastatic Relapse and Cause-specific Survival After Radical Treatment for Localised Prostate Cancer. *European Urology*, **2015**, 68, 186-93	10.2	192
883	Mosaic PPM1D mutations are associated with predisposition to breast and ovarian cancer. *Nature*, **2013**, 493, 406-10	50.4	191
882	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. *Nature Genetics*, **2017**, 49, 680-691	36.3	190
881	Germline RAD51C mutations confer susceptibility to ovarian cancer. *Nature Genetics*, **2012**, 44, 475-6; author reply 476	36.3	190
880	A new scoring system for the chances of identifying a BRCA1/2 mutation outperforms existing models including BRCAPRO. *Journal of Medical Genetics*, **2004**, 41, 474-80	5.8	189
879	Risk of cancer other than breast or ovarian in individuals with BRCA1 and BRCA2 mutations. *Familial Cancer*, **2012**, 11, 235-42	3	188
878	Management of the patient and family with neurofibromatosis 2: a consensus conference statement. *British Journal of Neurosurgery*, **2005**, 19, 5-12	1	187
877	Germline mutations in SUFU cause Gorlin syndrome-associated childhood medulloblastoma and redefine the risk associated with PTCH1 mutations. *Journal of Clinical Oncology*, **2014**, 32, 4155-61	2.2	185
876	Evaluation of clinical diagnostic criteria for neurofibromatosis 2. *Neurology*, **2002**, 59, 1759-65	6.5	185
875	Risk determination and prevention of breast cancer. *Breast Cancer Research*, **2014**, 16, 446	8.3	180
874	Predictors of the risk of mortality in neurofibromatosis 2. *American Journal of Human Genetics*, **2002**, 71, 715-23	11	178
873	Germline BRCA mutation and outcome in young-onset breast cancer (POSH): a prospective cohort study. *Lancet Oncology, The*, **2018**, 19, 169-180	21.7	177
872	A genetic register for von Hippel-Lindau disease. *Journal of Medical Genetics*, **1996**, 33, 120-7	5.8	169
871	Germ-line mutations of TP53 in Li-Fraumeni families: an extended study of 39 families. *Cancer Research*, **1997**, 57, 3245-52	10.1	169
870	Penetrance estimates for BRCA1 and BRCA2 based on genetic testing in a Clinical Cancer Genetics service setting: risks of breast/ovarian cancer quoted should reflect the cancer burden in the family. *BMC Cancer*, **2008**, 8, 155	4.8	165
869	Heritability of cellular radiosensitivity: a marker of low-penetrance predisposition genes in breast cancer?. *American Journal of Human Genetics*, **1999**, 65, 784-94	11	165
868	Prediction of pathogenic mutations in patients with early-onset breast cancer by family history. *Lancet, The*, **2003**, 361, 1101-2	40	164
867	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	8.1	164

866	Predictive testing for BRCA1/2: attributes, risk perception and management in a multi-centre clinical cohort. *British Journal of Cancer*, **2002**, 86, 1209-16	8.7	163
865	Loss-of-function mutations in SMARCE1 cause an inherited disorder of multiple spinal meningiomas. *Nature Genetics*, **2013**, 45, 295-8	36.3	162
864	Pregnancies, breast-feeding, and breast cancer risk in the International BRCA1/2 Carrier Cohort Study (IBCCS). *Journal of the National Cancer Institute*, **2006**, 98, 535-44	9.7	161
863	Cancer phenotype correlates with constitutional TP53 genotype in families with the Li-Fraumeni syndrome. *Oncogene*, **1998**, 17, 1061-8	9.2	160
862	A randomized placebo-controlled prevention trial of aspirin and/or resistant starch in young people with familial adenomatous polyposis. *Cancer Prevention Research*, **2011**, 4, 655-65	3.2	158
861	Targeted prostate cancer screening in BRCA1 and BRCA2 mutation carriers: results from the initial screening round of the IMPACT study. *European Urology*, **2014**, 66, 489-99	10.2	156
860	Paediatric presentation of type 2 neurofibromatosis. *Archives of Disease in Childhood*, **1999**, 81, 496-9	2.2	156
859	Women with neurofibromatosis 1 are at a moderately increased risk of developing breast cancer and should be considered for early screening. *Journal of Medical Genetics*, **2007**, 44, 481-4	5.8	155
858	The impact of genetic counselling on risk perception in women with a family history of breast cancer. *British Journal of Cancer*, **1994**, 70, 934-8	8.7	155
857	Frequent hSNF5/INI1 germline mutations in patients with rhabdoid tumor. *Clinical Cancer Research*, **2011**, 17, 31-8	12.9	153
856	Effect of chest X-rays on the risk of breast cancer among BRCA1/2 mutation carriers in the international BRCA1/2 carrier cohort study: a report from the EMBRACE, GENEPSO, GEO-HEBON, and IBCCS Collaborators' Group. *Journal of Clinical Oncology*, **2006**, 24, 3361-6	2.2	150
855	Common breast cancer susceptibility alleles and the risk of breast cancer for BRCA1 and BRCA2 mutation carriers: implications for risk prediction. *Cancer Research*, **2010**, 70, 9742-54	10.1	147
854	Perception of risk in women with a family history of breast cancer. *British Journal of Cancer*, **1993**, 67, 612-4	8.7	147
853	Exposure to diagnostic radiation and risk of breast cancer among carriers of BRCA1/2 mutations: retrospective cohort study (GENE-RAD-RISK). *BMJ, The*, **2012**, 345, e5660	5.9	145
852	Genotype/phenotype correlations in type 2 neurofibromatosis (NF2): evidence for more severe disease associated with truncating mutations. *Journal of Medical Genetics*, **1998**, 35, 450-5	5.8	145
851	Breast Cancer Risk Genes - Association Analysis in More than 113,000 Women. *New England Journal of Medicine*, **2021**, 384, 428-439	59.2	143
850	Molecular characterisation of SMARCB1 and NF2 in familial and sporadic schwannomatosis. *Journal of Medical Genetics*, **2008**, 45, 332-9	5.8	141
849	Whole-genome sequencing of patients with rare diseases in a national health system. *Nature*, **2020**, 583, 96-102	50.4	139

848	Mutational spectrum in a worldwide study of 29,700 families with BRCA1 or BRCA2 mutations. *Human Mutation*, **2018**, 39, 593-620	4.7	138
847	Cancer risk in Lynch Syndrome. *Familial Cancer*, **2013**, 12, 229-40	3	138
846	Screening for familial ovarian cancer: failure of current protocols to detect ovarian cancer at an early stage according to the international Federation of gynecology and obstetrics system. *Journal of Clinical Oncology*, **2005**, 23, 5588-96	2.2	134
845	Cancer Risks Associated With Germline Pathogenic Variants: An International Study of 524 Families. *Journal of Clinical Oncology*, **2020**, 38, 674-685	2.2	133
844	Predicting the likelihood of carrying a BRCA1 or BRCA2 mutation: validation of BOADICEA, BRCAPRO, IBIS, Myriad and the Manchester scoring system using data from UK genetics clinics. *Journal of Medical Genetics*, **2008**, 45, 425-31	5.8	129
843	Mortality in neurofibromatosis 1: in North West England: an assessment of actuarial survival in a region of the UK since 1989. *European Journal of Human Genetics*, **2011**, 19, 1187-91	5.3	128
842	Neurofibromatosis type 2. *Journal of Medical Genetics*, **2000**, 37, 897-904	5.8	128
841	Mammographic density adds accuracy to both the Tyrer-Cuzick and Gail breast cancer risk models in a prospective UK screening cohort. *Breast Cancer Research*, **2015**, 17, 147	8.3	124
840	Familial breast cancer. *Clinical Genetics*, **2012**, 82, 105-14	4	122
839	Increasing the specificity of diagnostic criteria for schwannomatosis. *Neurology*, **2006**, 66, 730-2	6.5	121
838	Genotype-phenotype correlations for nervous system tumors in neurofibromatosis 2: a population-based study. *American Journal of Human Genetics*, **2004**, 75, 231-9	11	121
837	A clinical study of type 1 neurofibromatosis in north west England. *Journal of Medical Genetics*, **1999**, 36, 197-203	5.8	121
836	Contralateral mastectomy improves survival in women with BRCA1/2-associated breast cancer. *Breast Cancer Research and Treatment*, **2013**, 140, 135-42	4.4	119
835	A genetic study of type 2 neurofibromatosis in the United Kingdom. II. Guidelines for genetic counselling. *Journal of Medical Genetics*, **1992**, 29, 847-52	5.8	117
834	Neurofibromatosis type 1 and autism spectrum disorder. *Pediatrics*, **2013**, 132, e1642-8	7.4	116
833	High resolution deletion analysis of constitutional DNA from neurofibromatosis type 2 (NF2) patients using microarray-CGH. *Human Molecular Genetics*, **2001**, 10, 271-82	5.6	115
832	Neurofibromatosis type 1 and sporadic optic gliomas. *Archives of Disease in Childhood*, **2002**, 87, 65-70	2.2	113
831	Autism and other psychiatric comorbidity in neurofibromatosis type 1: evidence from a population-based study. *Developmental Medicine and Child Neurology*, **2013**, 55, 139-45	3.3	111

830	Mammographic density and breast cancer risk in BRCA1 and BRCA2 mutation carriers. *Cancer Research*, **2006**, 66, 1866-72	10.1	111
829	Reproductive and hormonal factors, and ovarian cancer risk for BRCA1 and BRCA2 mutation carriers: results from the International BRCA1/2 Carrier Cohort Study. *Cancer Epidemiology Biomarkers and Prevention*, **2009**, 18, 601-10	4	110
828	BRCA1, BRCA2 and TP53 mutations in very early-onset breast cancer with associated risks to relatives. *European Journal of Cancer*, **2006**, 42, 1143-50	7.5	110
827	Prostate cancer in BRCA2 germline mutation carriers is associated with poorer prognosis. *British Journal of Cancer*, **2010**, 103, 918-24	8.7	109
826	Uptake of risk-reducing surgery in unaffected women at high risk of breast and ovarian cancer is risk, age, and time dependent. *Cancer Epidemiology Biomarkers and Prevention*, **2009**, 18, 2318-24	4	109
825	The gene for the naevoid basal cell carcinoma syndrome acts as a tumour-suppressor gene in medulloblastoma. *British Journal of Cancer*, **1997**, 76, 141-5	8.7	108
824	Familial infiltrative fibromatosis (desmoid tumours) (MIM135290) caused by a recurrent 3' APC gene mutation. *Human Molecular Genetics*, **1996**, 5, 1921-4	5.6	108
823	Baseline Surveillance in Li-Fraumeni Syndrome Using Whole-Body Magnetic Resonance Imaging: A Meta-analysis. *JAMA Oncology*, **2017**, 3, 1634-1639	13.4	107
822	Mosaicism in neurofibromatosis type 2: an update of risk based on uni/bilaterality of vestibular schwannoma at presentation and sensitive mutation analysis including multiple ligation-dependent probe amplification. *Journal of Medical Genetics*, **2007**, 44, 424-8	5.8	107
821	Breast and ovarian cancer risks to carriers of the BRCA1 5382insC and 185delAG and BRCA2 6174delT mutations: a combined analysis of 22 population based studies. *Journal of Medical Genetics*, **2005**, 42, 602-3	5.8	107
820	Germline mutation of ARF in a melanoma kindred. *Human Molecular Genetics*, **2002**, 11, 1273-9	5.6	107
819	Germline selection shapes human mitochondrial DNA diversity. *Science*, **2019**, 364,	33.3	105
818	Evidence of Stage Shift in Women Diagnosed With Ovarian Cancer During Phase II of the United Kingdom Familial Ovarian Cancer Screening Study. *Journal of Clinical Oncology*, **2017**, 35, 1411-1420	2.2	105
817	MRI breast screening in high-risk women: cancer detection and survival analysis. *Breast Cancer Research and Treatment*, **2014**, 145, 663-72	4.4	105
816	Assessment of in vitro sperm characteristics in relation to fertility in dairy bulls. *Animal Reproduction Science*, **2008**, 103, 201-14	2.1	104
815	Variants in CHEK2 other than 1100delC do not make a major contribution to breast cancer susceptibility. *American Journal of Human Genetics*, **2003**, 72, 1023-8	11	104
814	Loss of SUFU function in familial multiple meningioma. *American Journal of Human Genetics*, **2012**, 91, 520-6	11	103
813	Results of annual screening in phase I of the United Kingdom familial ovarian cancer screening study highlight the need for strict adherence to screening schedule. *Journal of Clinical Oncology*, **2013**, 31, 49-57	2.2	103

812	Screening for familial ovarian cancer: poor survival of BRCA1/2 related cancers. *Journal of Medical Genetics*, **2009**, 46, 593-7	5.8	103
811	Somatic mosaicism in neurofibromatosis 2: prevalence and risk of disease transmission to offspring. *Journal of Medical Genetics*, **2003**, 40, 459-63	5.8	103
810	Clinical follow-up after bilateral risk reducing ('prophylactic') mastectomy: mental health and body image outcomes. *Psycho-Oncology*, **2000**, 9, 462-72	3.9	103
809	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	2.2	101
808	Neurofibromatosis 2, radiosurgery and malignant nervous system tumours. *British Journal of Cancer* , **2000**, 82, 998	8.7	101
807	A genome wide linkage search for breast cancer susceptibility genes. *Genes Chromosomes and Cancer*, **2006**, 45, 646-55	5	100
806	Germline mutations in the neurofibromatosis type 2 tumour suppressor gene. *Human Molecular Genetics*, **1994**, 3, 813-6	5.6	100
805	Bilateral Oophorectomy and Breast Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. *Journal of the National Cancer Institute*, **2017**, 109,	9.7	99
804	Impaired tamoxifen metabolism reduces survival in familial breast cancer patients. *Clinical Cancer Research*, **2008**, 14, 5913-8	12.9	99
803	Life expectancy in hereditary cancer predisposing diseases: an observational study. *Journal of Medical Genetics*, **2012**, 49, 264-9	5.8	98
802	Neurofibromatosis 2 [Bilateral acoustic neurofibromatosis, central neurofibromatosis, NF2, neurofibromatosis type II]. *Genetics in Medicine*, **2009**, 11, 599-610	8.1	98
801	Current policies for surveillance and management in women at risk of breast and ovarian cancer: a survey among 16 European family cancer clinics. European Familial Breast Cancer Collaborative Group. *European Journal of Cancer*, **1998**, 34, 1922-6	7.5	98
800	Cancer Surveillance in Gorlin Syndrome and Rhabdoid Tumor Predisposition Syndrome. *Clinical Cancer Research*, **2017**, 23, e62-e67	12.9	97
799	Cancer and Central Nervous System Tumor Surveillance in Pediatric Neurofibromatosis 1. *Clinical Cancer Research*, **2017**, 23, e46-e53	12.9	95
798	Analysis of genetic and phenotypic heterogeneity in juvenile polyposis. *Gut*, **2000**, 46, 656-60	19.2	95
797	The MDM2 promoter SNP285C/309G haplotype diminishes Sp1 transcription factor binding and reduces risk for breast and ovarian cancer in Caucasians. *Cancer Cell*, **2011**, 19, 273-82	24.3	94
796	Cost-effectiveness of screening with contrast enhanced magnetic resonance imaging vs X-ray mammography of women at a high familial risk of breast cancer. *British Journal of Cancer*, **2006**, 95, 801-10	8.7	94
795	Genotype-Phenotype Correlation in NF1: Evidence for a More Severe Phenotype Associated with Missense Mutations Affecting NF1 Codons 844-848. *American Journal of Human Genetics*, **2018**, 102, 69-87	11	93

794	Germline and somatic NF1 gene mutation spectrum in NF1-associated malignant peripheral nerve sheath tumors (MPNSTs). *Human Mutation*, **2008**, 29, 74-82	4.7	93
793	Neurofibromatosis 2. *Current Opinion in Neurology*, **2003**, 16, 27-33	7.1	93
792	Magnetic resonance imaging screening in women at genetic risk of breast cancer: imaging and analysis protocol for the UK multicentre study. UK MRI Breast Screening Study Advisory Group. *Magnetic Resonance Imaging*, **2000**, 18, 765-76	3.3	92
791	Common variants in LSP1, 2q35 and 8q24 and breast cancer risk for BRCA1 and BRCA2 mutation carriers. *Human Molecular Genetics*, **2009**, 18, 4442-56	5.6	91
790	Phenocopies in BRCA1 and BRCA2 families: evidence for modifier genes and implications for screening. *Journal of Medical Genetics*, **2007**, 44, 10-15	5.8	91
789	Survival in prospectively ascertained familial breast cancer: analysis of a series stratified by tumour characteristics, BRCA mutations and oophorectomy. *International Journal of Cancer*, **2002**, 101, 555-9	7.5	91
788	Risk perception and cancer worry: an exploratory study of the impact of genetic risk counselling in women with a family history of breast cancer. *Journal of Medical Genetics*, **2001**, 38, 139	5.8	91
787	Cancer risk and genotype-phenotype correlations in PTEN hamartoma tumor syndrome. *Familial Cancer*, **2014**, 13, 57-63	3	90
786	Identification of a BRCA2-specific modifier locus at 6p24 related to breast cancer risk. *PLoS Genetics*, **2013**, 9, e1003173	6	90
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